

Women's Health Partners, LLC

Diplomates American Board of Obstetrics & Gynecology

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Carrier Testing for Common Genetic Diseases

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

There are a few common inherited diseases that can occur even without a family history and that can be tested for in the general population. Four of these are cystic fibrosis (CF), spinal muscular atrophy (SMA), fragile X syndrome, and hemoglobinopathies. A simple blood test on a sample of your blood can help to determine if you are a carrier for one of these diseases.

If you still have any questions or concerns, we strongly encourage you to contact our office prior to your procedure 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. 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 the family history with your doctor or a genetics 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 CF, SMA, or a hemoglobinopathy, the next step is for your partner to have carrier testing performed. 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.

If the test shows that you are a carrier of fragile X, your partner does not need testing because this disease is usually inherited only through the woman. Therefore, the next step is for you to consider prenatal testing by amniocentesis or chorionic villi sampling (CVS) to determine if your baby is affected.

Insurance Coverage

These tests may or may not be covered by your health insurance company. The lab company will bill your insurance directly 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 them if you want them to proceed with the test or not. If not covered, they will offer you a cash price. If you have questions contact your insurance company for more information.

- I want CF, SMA, fragile X, hemoglobinopathy carrier testing.
- I want carrier testing for: cystic fibrosis SMA fragile X hemoglobinopathies
- I do not want CF, SMA, fragile X, hemoglobinopathies carrier testing.

Patient Signature

Date

Account #

Patient Name (Print)

Witness

Date

Cystic Fibrosis (CF)	Spinal Muscular Atrophy (SMA)	Fragile X Syndrome	Hemoglobinopathies
<p>The most common inherited disease of children and young adults.</p> <p>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.</p>	<p>The most common inherited cause of early childhood death.</p> <p>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.</p>	<p>The most common inherited cause of mental retardation.</p> <p>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.</p>	<p>Hemoglobinopathies are one of the most common inherited disorders of in the world.</p> <p>Alpha thalassemia is a blood disorder that reduces the production of hemoglobin.</p> <p>Beta hemoglobinopathies include sickle cell anemia and beta thalassemia which can cause disease.</p>
<p>Inheritance:</p> <p>If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.</p>	<p>Inheritance:</p> <p>If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA.</p>	<p>Inheritance:</p> <p>If a mother is a carrier, there is a 50% chance to have a child with fragile X syndrome.</p>	<p>Inheritance:</p> <p>Alpha thalassemia results from mutations in the alpha-globin genes.</p> <p>Beta hemoglobin problems result from mutations in the beta globin genes.</p>
<p>Carrier Frequency:</p> <p>1 in 30 average in the U.S. Varies by ethnicity</p>	<p>Carrier Frequency:</p> <p>1 in 54 in all ethnicities.</p>	<p>Carrier Frequency:</p> <p>~1 in 260 women Occurs in all ethnic backgrounds</p>	<p>Carrier Frequency:</p> <p>The carrier frequency of hemoglobinopathies varies by the type and by the population evaluated.</p>
<p>For CF:</p> <p>If I am a carrier, testing my partner will help me learn more about the chance that my baby could have CF.</p> <p>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%.</p> <p>If both parents are carriers, prenatal testing is available to find out whether my baby has inherited the abnormal CF genes.</p>	<p>For SMA:</p> <p>If I am a carrier, testing my partner will help me learn more about the chance that my baby could have SMA.</p> <p>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%.</p> <p>If both parents are carriers, prenatal testing is available to find out whether the baby has inherited the abnormal SMA genes.</p>	<p>For Fragile X:</p> <p>If I am a carrier, prenatal testing is available to find out whether my baby has inherited the abnormal fragile X gene.</p>	<p>For hemoglobinopathies:</p> <p>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.</p>
<p>You should be certain you understand the following points:</p> <ul style="list-style-type: none"> <input type="checkbox"/> The purpose of these tests is to determine whether I am a carrier of one of the common genetic abnormalities that cause CF, SMA and/or fragile X syndrome. <input type="checkbox"/> The tests do not detect all carriers of these diseases. <input type="checkbox"/> The laboratory needs accurate information about my family history for the most accurate interpretation of the test results. <input type="checkbox"/> The decision to have carrier testing is completely mine. <p>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 genetics professional. I have all the information I want, and all my questions have been answered.</p>			