

# Carrier Testing for Common Genetic Diseases



**Cystic Fibrosis**  
**Spinal Muscular Atrophy**  
**Fragile X Syndrome**



## Carrier Testing for Common Genetic Diseases

This brochure provides information to help you decide about having carrier testing for three common genetic diseases. If you have any additional questions please talk to your doctor or to a genetics 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. Three of these are cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome. A simple blood test on a sample of your blood can help to determine if you are a carrier for one of these diseases.

### What are these common genetic diseases?

The chart on the back provides a summary of information about CF, SMA and fragile X syndrome, and carrier screening for these diseases.

### 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, and, 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.

### If your test is negative, could you still be a carrier?

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.

### What if the test shows you are a carrier of CF, SMA or fragile X?

If the test shows that you are a carrier of CF, 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 CF. If your partner has a negative test result and no family history of CF, the chance that your baby will have CF 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 SMA, 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 SMA. If your partner has a negative test result and no family history of SMA, the chance that your baby will have SMA 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.

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Disease	Cystic Fibrosis (CF)	Spinal Muscular Atrophy (SMA)	Fragile X Syndrome
<b>Symptoms of Disease</b>	<p><b>The most common inherited disease of children and young adults.</b></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><b>The most common inherited cause of early childhood death.</b></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><b>The most common inherited cause of mental retardation.</b></p> <p>Fragile X syndrome involves developmental delay, mental retardation, autism and hyperactivity. It primarily affects boys. Women who are carriers are at risk to have a child with mental retardation.</p>
<b>Inheritance</b>	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA.	If a mother is a carrier, there is a 50% chance to have a child with fragile X syndrome.
<b>General Population Carrier Frequency</b>	1 in 30 average in the U.S. Varies by ethnicity	Ranges from 1 in 47 to 1 in 72 in the U.S. Varies by ethnicity	~1 in 260 women Occurs in all ethnic backgrounds
<b>Model Informed Consent/Decline for Carrier Testing</b>	<p><b>You should be certain you understand the following points:</b></p> <ul style="list-style-type: none"> <li>■ 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.</li> <li>■ The tests do not detect all carriers of these diseases.</li> <li>■ The laboratory needs accurate information about my family history for the most accurate interpretation of the test results.</li> <li>■ The decision to have carrier testing is completely mine.</li> <li>■ No other test will be performed and reported on my sample unless authorized by my doctor, and any unused portion of my original sample will be destroyed within two months of receipt of the sample by the laboratory.</li> <li>■ The laboratory will disclose the test results ONLY to my doctor, or to his/her agent, unless otherwise authorized by me or required by law.</li> </ul>		
	<p><b>For CF:</b></p> <ul style="list-style-type: none"> <li>■ If I am a carrier, testing my partner will help me learn more about the chance that our baby could have CF.</li> <li>■ 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%.</li> <li>■ If both parents are carriers, prenatal testing is available to find out whether or not the baby has inherited the abnormal CF genes.</li> </ul>	<p><b>For SMA:</b></p> <ul style="list-style-type: none"> <li>■ If I am a carrier, testing my partner will help me learn more about the chance that our baby could have SMA.</li> <li>■ 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%.</li> <li>■ If both parents are carriers, prenatal testing is available to find out whether or not the baby has inherited the abnormal SMA genes.</li> </ul>	<p><b>For Fragile X:</b></p> <ul style="list-style-type: none"> <li>■ If I am a carrier, prenatal testing is available to find out whether or not the baby has inherited the abnormal fragile X gene.</li> </ul>
	<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. I have decided that:</p>		
	<p><input type="checkbox"/> I want CF carrier testing.</p> <p><input type="checkbox"/> I do not want CF carrier testing.</p> <p><b>Patient Signature:</b> _____</p> <p><b>Date:</b> _____</p>	<p><input type="checkbox"/> I want SMA carrier testing.</p> <p><input type="checkbox"/> I do not want SMA carrier testing.</p> <p><b>Patient Signature:</b> _____</p> <p><b>Date:</b> _____</p>	<p><input type="checkbox"/> I want fragile X carrier testing.</p> <p><input type="checkbox"/> I do not want fragile X carrier testing.</p> <p><b>Patient Signature:</b> _____</p> <p><b>Date:</b> _____</p>

#### About Integrated Genetics

Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years. This brochure is provided by Integrated Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites: [www.mytestingoptions.com](http://www.mytestingoptions.com) and [www.integratedgenetics.com](http://www.integratedgenetics.com).

