



CLAIRE ALTMAN HEINE FOUNDATION, INC.

dedicated to identifying carriers of SMA

Carrier Testing

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What Is Carrier Testing?

Carrier testing is genetic testing to determine whether an individual carries one copy of an altered gene for a particular recessive condition. Genes are the basic units of heredity passed from parent to child. Genes occur in pairs and we inherit one member of each pair from our mother and the other from our father. A change in a gene, known as a mutation, can cause that gene not to work properly. This can lead to disease.

Certain diseases are caused when both genes of a pair have mutations. This is called a recessive disease. In order to have a child with a recessive disease, each parent has a gene that works properly and a gene that does not. The parents are known as carriers and do not have any health problems related to that gene. Genetic testing can identify carriers by a simple blood test to determine if a couple is at risk to have a child affected with a specific disease.

What is the Purpose of SMA Carrier Testing?

The purpose of SMA carrier testing is to see if a couple is at an increased risk of giving birth to a child who will have SMA. SMA carrier testing is a laboratory test done on a sample of blood. If testing shows that a couple is at high risk, additional testing can be done during pregnancy to see whether or not the baby will have SMA. However, most women's test results are normal.

What are the Benefits of Carrier Testing?

Carrier testing can help couples make decisions about whether and how to have children. Couples have many options. Some couples may choose not to risk having a child born with SMA and may adopt, use donated eggs or sperm or go through in vitro fertilization and test the embryos using preimplantation genetic diagnosis (PGD). Other couples may choose to become pregnant and pursue the earliest available prenatal testing. Some couples may use carrier testing to learn about their risks before they become pregnant but not pursue prenatal testing.

Could I be a carrier of SMA?

Yes. You could be a carrier of SMA even if no one in your family has SMA and even if you already have children without SMA. Carriers of the abnormal gene for SMA have no symptoms and are not sick. About one of every 40 people carries the gene. If a relative of yours has SMA, or is known to be a carrier of SMA, your chance of being a carrier is greater based on your family history.

If my test result is normal could I still be a carrier?

Yes. However, this is rare. The likelihood that you are a carrier even though you had a normal result is small.

If the test shows I am a carrier what should I do?

If the test shows that you are a carrier, you may want to test the baby's father. Both parents must be carriers for the baby to have SMA. If the father has a normal test result and no family history of SMA, the chance that your baby will have SMA is very, very small. Since this is a very rare occurrence, if you are a carrier but the father has a normal result, no further testing would be recommended.

What happens if both my partner and I are carriers?

What if both my partner and I are SMA carriers?

The test results mean that you and your partner are both carriers of the abnormal SMA gene. This does not have any impact on your health. However, it means that you are at risk for having a baby with SMA. "It is important to remember that the result you received does not mean that your baby will definitely have SMA.

If you and your partner have both been shown to be carriers of an abnormal SMA gene and have a child together, there is a 1-in-4 (25%) chance with each pregnancy that the child will have SMA. This is true even if you already have other children with or without SMA.

If SMA testing shows both parents are carriers, you might see a genetic counselor. There are several choices couples in your situation can make when thinking about possible future pregnancies. Some couples decided to:

- Accept this level of risk and have children without further testing.
- Have prenatal testing, such as amniocentesis or CVS to determine if the fetus has SMA.
- Use donor sperm or donor eggs.
- Adopt.
- Not have other children.

What is the chance that my baby will have SMA?

There is a 1-in-4 (25%) chance that the baby will have SMA. Stated another way, there is a 3-in-4 (75%) chance that your baby will not have SMA.

There is a 1-in-2 (50%) chance that your baby will be a carrier, like you and your partner. This will not have health consequences for the baby, but she or he could be a risk for having a child with SMA in the future.

There is also a 1-in-4 (25%) chance that the baby will not have the changed SMA gene - that is, neither have the disease nor be able to pass on the gene.

Can I find out if my baby will have SMA?

Yes, through additional testing during pregnancy. Chorionic villus sampling (CVS), generally done around the 11th week of pregnancy, is done by using a small instrument to remove a very small portion of the placenta. This portion is analyzed to determine if the baby has SMA. Alternatively amniocentesis, generally done around the 16th week of pregnancy, is performed by removing a small amount of fluid surrounding the baby using a thin needle inserted into a woman's uterus. The fluid is analyzed to determine if the baby has SMA.

If you decide not to have this prenatal testing, you can find out whether or not the baby is affected with SMA after its birth through a simple test done with a blood sample.

Are the results of the tests definite?

Yes. The results of these prenatal tests (that is amniocentesis or CVS) are nearly 100% accurate about whether or not the baby will have SMA. However, they usually cannot tell you how severe or mild the disease will be if the baby has SMA.

Might others in my family be SMA carriers?

Yes. Even if no one in your family has had SMA, other close relatives, such as brothers, sisters, aunts, uncles and cousins, may also be carriers. This information might be useful for them to know if they are planning pregnancies. This is something you might want to discuss further with your care provider.

What if the amniocentesis or CVS results show that the baby does not have SMA?

If the test results indicate the baby will not have SMA, you can go through the rest of your pregnancy feeling assured that the baby will not develop this condition. There is a 3-in-4 (75%) chance that this will be the result of your prenatal test.

What about future pregnancies?

It is important to remember that you and your partner have both been shown to be carriers of an abnormal SMA gene. This means that in each pregnancy the two of you have together, that baby will also have a 1-in-4 (25%) chance of having SMA. If you want to know whether or not that baby will develop SMA, you need to have amniocentesis or CVS in each pregnancy.

There are several choices couples in your situation can make when thinking about possible future pregnancies. Some couples decide to:

- Have prenatal testing, such as amniocentesis or CVS
- Accept this level of risk and have children without further testing
- Go through in vitro fertilization and test the embryos using Preimplantation genetic Diagnosis (PGD)
- Adopt
- Use donor sperm or donor eggs
- Not have other children

If all tests show that the baby will have SMA, what are my options?

SMA is not a curable disease. There are no known treatments available. Therefore, there are only three decisions that are possible once it is found out that a fetus has SMA.

You can continue the pregnancy and prepare for the addition to your family of a child with SMA. Couples can use this time to learn as much as possible about the disease by talking with care providers and the experiences of other families who have a child or children with SMA.

You may choose to terminate the pregnancy. If this is an option that you might consider, you should discuss with your doctor. This must usually be done before the end of the second trimester of pregnancy. Each state has somewhat different laws on pregnancy termination.

You may choose to continue the pregnancy and prepare to place the child up for adoption.