Spinal Muscular Atrophy

The most common inherited cause of early childhood death

Integrated GENETICS

LabCorp Specialty Testing Group

Spinal Muscular Atrophy Resources

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Thanks to the Claire Altman Heine Foundation for providing content for this spinal muscular atrophy patient guide.

Informed Consent/Decline for SMA Carrier Screening

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My signature below indicates that I have read, or had read to me, the above information and I understand it. I have also read or had explained to me the specific disease(s) or condition(s) tested for, and the specific test(s) I am having, including the test descriptions, principles and limitations.

I have had the opportunity to discuss the purposes and possible risks of this testing with my doctor or someone my doctor has designated. I know that genetic counseling is available to me before and after the testing. I have all the information I want, and all my questions have been answered.

I have decided that:

☐ **I want SMA carrier testing.**

☐ **I do not want SMA carrier testing.**

Patient Signature

Date

Obtained by

This model informed consent form is provided by Integrated Genetics as a courtesy to physicians and their patients.

Informed Consent/Decline for Spinal Muscular Atrophy (SMA) Carrier Screening

You should be certain you understand the following points:

1. The purpose of my genetic testing is to determine whether I, or my baby, have mutation(s) known to be associated with SMA.
2. This testing is done on a small sample of blood. For the baby, testing is done on amniotic fluid, CVS, or fetal blood.
3. Mutations are often different in different populations. I understand that the laboratory needs accurate information about my family history and ethnic background for the most accurate interpretation of the test results.
4. When SMA testing shows a mutation, then the person is a carrier or is affected with the condition or disease tested for. Consulting a doctor or genetic counselor is recommended to learn the full meaning of the results and to learn if additional testing might be necessary.
5. When the SMA testing does not show a known mutation, the chance that the person is a carrier or is affected is reduced. There is still a chance to be a carrier or to be affected because the current testing cannot find all the possible changes within a gene.
6. In some families, SMA testing might discover non-paternity (someone who is not the real father), or some other previously unknown information about family relationships, such as adoption.
7. In the case of twins or other multiple babies, the results may pertain to only one of the babies.
8. In the case of abnormal diagnostic results, the decision to continue or terminate the pregnancy is entirely mine.
9. The decision to consent to or to refuse SMA testing is entirely mine.
10. No test(s) will be performed and reported on my sample other than those authorized by my doctor; and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the laboratory.
11. My doctor may release my pregnancy outcome or ultrasound and amniocentesis results to Esoterix Genetic Laboratories, LLC. to be used for statistical analysis of the laboratory's performance.
12. Esoterix Genetic Laboratories, LLC. will disclose the test results only to my doctor or to his/her agent unless otherwise authorized by me or required by law.

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If my test is negative, could I still be a carrier?

A negative result significantly lowers, but does not completely eliminate, the risk of being a carrier of SMA. Carrier screening does not detect less common abnormalities (mutations) that cause SMA.

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

If the test determines that you are a carrier, 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%.

What if both my partner and I are SMA carriers?

It is important to remember that if you and your partner are both found 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 be affected with SMA. This is true even if you already have other children with or without SMA.

If SMA screening shows both parents are carriers, you may be referred to a genetic counselor. 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, to determine whether or not the unborn baby has inherited the two abnormal SMA genes
- 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 children
- Use donor sperm or donor eggs
- Not have children

- 50% (2 out of 4) chance that the child will inherit one normal and one abnormal SMA gene, and will be a carrier of SMA (but not affected)
- 25% (1 out of 4) chance that the child will inherit two abnormal SMA genes – one from each parent – and will be affected with the disease

Could I be a carrier of SMA?

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 have no symptoms of the disease. As with most inherited diseases, the risk for being an SMA carrier varies by ethnic background. Because SMA is such a severe disease with a high carrier frequency, ACMG guidelines recommend all people be offered testing regardless of race or ethnicity.[1]

SMA Carrier Risk in People with No Family History of SMA:[3]

- Caucasian 1 in 47
- Asian Indian 1 in 52
- Asian 1 in 59
- Ashkenazi Jewish 1 in 67
- Hispanic 1 in 68
- African American 1 in 72

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.

What is the purpose of SMA carrier screening?

The purpose of SMA carrier screening is to see if a couple is at increased risk for giving birth to a child who will be affected with SMA. The test is done on a sample of blood. If results show that a couple is at high risk, prenatal testing (chorionic villi sampling [CVS] or amniocentesis) during pregnancy or preimplantation testing before pregnancy can be done to see whether or not the baby has inherited two abnormal SMA genes. Neither carrier screening nor prenatal diagnostic testing can tell what type of SMA the child could have.

Spinal Muscular Atrophy (SMA)

This brochure contains general information about Spinal Muscular Atrophy (SMA) carrier screening and how SMA is inherited. It is our hope that you find this information helpful, but if you have any additional questions please contact your doctor or a genetics professional.

Given the severity and frequency of this disease, the American College of Medical Genetics (ACMG) recommends SMA carrier screening be offered before conception or early in pregnancy to everyone. The primary goal is to allow carriers to make informed reproductive choices.[1]

What is SMA?

SMA is a hereditary disease that destroys the nerves responsible for controlling voluntary muscle movement, but does not affect intelligence. Muscles that control breathing, swallowing, head and neck control, walking and crawling are the most severely affected. SMA is a variable disease in terms of when the symptoms begin. Most often it shows up before a baby is two years old, but in some individuals the symptoms start before birth and in others not until after age 30. Over 60% of individuals who are diagnosed with SMA are severely affected.[2] There is currently no cure or treatment for SMA.

How is SMA inherited?

If both parents are carriers of an abnormal SMA gene, there is a chance that each parent will pass the abnormal gene on to their child. An individual with two abnormal SMA genes will be affected with the disease. With each pregnancy there is a:

- 25% (1 out of 4) chance that the child will inherit two normal SMA genes, and will be neither a carrier nor affected with the disease

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
www.integratedgenetics.com

References:

1) Prior TW. ACMG Practice Guidelines: Carrier screening for spinal muscular atrophy. *Genet Med* 2008; 10:840-842.
2) Meldrum C, et al. Spinal muscular atrophy genetic counseling access and genetic knowledge: Parents' Perspectives. *J Child Neurol* 2007; 22:1019-1026.
3) Sugarman EA, et al. Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. *Eur J Hum Genet* 2012; 20:27-32.

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