Consent form

Informed consent/decline for spinal muscular atrophy (SMA) carrier screening

My signature below indicates that I have read, or had read to me, the provided information and I understand it. I have also read or had explained to me the specific disorder(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 screening.
- ☐ I do not want SMA carrier screening.

After completing this form, tear off and provide back to your physician or provider.

Patient Signature

Date

Obtained by

This consent form is provided by Labcorp as a courtesy to physicians and their patients. For additional information and the model informed consent form, visit womenshealth.labcorp.com

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Pioneering science, personalized service

Convenient blood draws
Getting your blood drawn is easier than ever. Labcorp has a nationwide network of patient service centers, allowing for convenient access to sample collection. Visit labcorp.com to find your nearest location.

Genetic counseling
Patients with a positive test result may be offered counseling, and Labcorp offers the largest national commercial network of genetic counselors to help inform and support patients.

General questions?
Call toll-free within the U.S.
(800) 848-4436

References
1. Carrier Screening for Genetic Conditions. Committee Opinion No. 691. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2017;129:e41–55

2. Gregg AR, Aarabi M, Klugman S, et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med.* 2021;23(10):1793-1806. doi:10.1038/s41436-021-01203-z.

3. Meldrum C, et al. Spinal muscular atrophy genetic counseling access and genetic knowledge: Parents' Perspectives. *J Child Neurol* 2007; 22:1019-1026.

4. 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.

Note: This material is provided for general information purposes only. It is not intended as a substitute for medical advice and/or consultation with a physician or technical expert.

PATIENT INFORMATION

Spinal Muscular Atrophy

The most common inherited cause of early childhood mortality. Earlier diagnosis can lead to better outcomes.

Carrier screening for spinal muscular atrophy (SMA)

Professional societies recommend SMA carrier screening be offered to all patients who are currently pregnant or considering pregnancy. Screening can help determine if you are at increased risk of having a child with SMA. Early insight can help you better prepare for your family's future. [1,2]

What is SMA?
SMA is a hereditary disorder 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 disorder in terms of when the symptoms begin. Symptoms often appear 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.[3]

How is SMA inherited?
When both biological parents are carriers of a non-functional SMA gene, there is a chance that each will pass that gene onto future offspring. An individual with two non-functional SMA genes will have the disorder. With each pregnancy there is a:

- 25% (1 out of 4) chance that future offspring will inherit two normal SMA genes and will be neither a carrier nor affected
- 50% (2 out of 4) chance that future offspring will inherit one normal and one non-functional SMA gene and will be a carrier of SMA (but not affected)
- 25% (1 out of 4) chance that future offspring will inherit two non-functional SMA genes, one from each parent, and will be affected with the disorder

What is the treatment for SMA?
There are currently several FDA-approved treatments for SMA. Timing can be critical for the effectiveness of these therapies. Early detection can lead to enhanced neonatal care and improved outcomes.

Can anyone be an SMA carrier?
Yes. SMA can occur in any ethnic background. 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 non-functional gene have no symptoms of the disorder. As with most inherited disorders, the risk for being an SMA carrier varies by ethnic background. Because SMA is such a severe disorder with a high carrier frequency, professional societies recommend all people be offered testing regardless of race or ethnicity.[1]

SMA carrier risk in people with no family history of SMA:[4]	
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 the biological parents are at increased risk to have future offspring with SMA. The test is performed on a sample of blood or saliva. If results show a high risk, prenatal testing (chorionic villi sampling [CVS] or amniocentesis) during pregnancy or preimplantation testing (PGT-M) before pregnancy can be done to see whether or not two non-functional SMA genes have been inherited. Neither carrier screening nor prenatal diagnostic testing can tell what type of SMA a child could have.

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 (pathogenic variants) 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 biological parents must be carriers for future offspring to be at risk for SMA. If one biological partner has a negative test result and no family history of SMA, the chance future offspring 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 biological partner are both found to be carriers of a non-functional SMA gene, there is a 1-in-4 (25%) chance with each pregnancy that future offspring will be affected with SMA. This is true even if you already have other children with or without SMA.

If SMA screening shows both biological parents are carriers, you may be referred to a genetic counselor. There are several choices you can make when thinking about possible future pregnancies. You may decide to:

- Have prenatal testing, such as amniocentesis or CVS, to determine whether or not the unborn baby has inherited the two non-functional SMA genes
- Accept this level of risk and have children without further testing
- Pursue in vitro fertilization and test the embryos using preimplantation genetic testing (PGT-M)
- Adopt children
- Use donor sperm or donor eggs
- Choose to live child-free

Spinal muscular atrophy resources

Cure SMA
925 Busse Road
Elk Grove Village, IL 60007
(800) 886-1762
curesma.org

National Society of Genetic Counselors
401 N. Michigan Avenue
Chicago, IL 60611
(312) 321-6834
nsgc.org

Genetic Alliance
4301 Connecticut Avenue NW, Suite 404
Washington, D.C. 20008-2369
(202) 966-5557
geneticalliance.org

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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 pathogenic variant(s) known to be associated with SMA.
2. This testing is done on a small sample of blood or saliva. Fetal testing is done on amniotic fluid, chorionic villi or fetal blood.
3. I understand 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 non-functional gene(s), then the person is a carrier or is affected with the condition. 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 non-functional gene(s), 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 biological 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 Labcorp to be used for statistical analysis of the laboratory's performance.
12. Labcorp will disclose the test results only to me and my doctor or to his/her agent unless otherwise authorized by me or required by law.
13. If I am a carrier, additional testing will be offered to help determine reproductive risk.

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