ACOG
The American College of Obstetricians and Gynecologists

FAQs

Carrier Screening for Spinal Muscular Atrophy (SMA)

Frequently Asked Questions

What is spinal muscular atrophy?

Spinal muscular atrophy (SMA) is a genetic disorder that affects the nerves of the spine. These nerves control muscles for breathing, swallowing, and movement of the arms and legs. SMA causes these muscles to atrophy (get smaller) and become very weak. Depending on the type, SMA can cause severe disability and death. SMA does not affect mental ability.

What are the different types of spinal muscular atrophy?

There are four types of SMA. Babies born with type I, the most common type, may be severely disabled, cannot sit or stand, and usually die before age 2 years. Children with type II need help walking and standing and have a shortened lifespan. Children with type III are less disabled and may be able to stand on their own, sometimes without help. Type IV begins in adulthood.

What causes spinal muscular atrophy?

SMA is caused by changes (called mutations) in a gene called SMN1. Genes are the instructions that control a function in the body or a physical trait, like eye color. A person

with SMA has two faulty copies of SMN1, one from their father and one from their mother.

Can spinal muscular atrophy be prevented?

No, SMA cannot be prevented and there is no cure.

What does it mean to be a carrier of spinal muscular atrophy?

A carrier is a person who inherits one healthy copy and one faulty copy of the SMN1 gene. About 1 in 40 to 1 in 60 people are carriers of SMA. If both parents are carriers, they have a 1-in-4 chance of having a child with SMA. About 1 in 6,000 to 1 in 10,000 children are born with SMA.

My family member has spinal muscular atrophy. Does this increase my risk of having a child with the disorder?

Yes. SMA is an inherited disorder that runs in families. If you have a family member who has SMA, it means that your risk of being a carrier is increased.

What is carrier screening?

Carrier screening is a type of test that can tell whether you carry a change in a gene for certain genetic disorders. When screening is done before or during pregnancy, it allows you to find out the chances of having a child with a genetic disorder.

Carrier screening for SMA can tell you whether you are at risk of having a baby with SMA, but it cannot tell you with 100% certainty. Also, if screening shows that both parents are carriers, the results cannot tell you how severe the disease may be if one of your children inherits it.

How is carrier screening done?

Carrier screening is done by testing a sample of blood. One parent usually is tested first. If results show that the first parent is a carrier, the other parent is tested.

What do carrier screening results mean?

Carrier screening results for SMA are reported as the number of healthy copies of *SMN1* a person has:

- If you have two copies of the healthy gene, it means that you have a reduced risk of being a carrier.
- If you have one healthy copy of *SMN1*, it means the other copy is faulty and you are a carrier. You could pass the faulty copy of the *SMN1* gene to your child.

What happens if test results show I am a carrier?

The next step is to have your partner tested. If your partner also is a carrier, your risk of having a child with SMA is 1 in 4. Your risk of having a child who is a carrier is 1 in 2. If your partner has two healthy copies of the SMN1 gene, your risk of having a child with the disorder is very low.

Who should have carrier screening?

All women who are thinking about getting pregnant or who are already pregnant should be offered carrier screening for SMA. You can have screening for additional disorders as well. You can choose to have carrier screening, or you can choose not to. There is no right or wrong choice.

When should I have carrier screening?

Carrier screening can be done either before pregnancy or during pregnancy. Having carrier screening before you get pregnant gives you more options than having it during pregnancy.

My partner and I are both carriers. What are our options for pregnancy?

If you find out that you and your partner are carriers before pregnancy, you have the following choices:

- You can get pregnant and then have amniocentesis or chorionic villus sampling (CVS) to see if the fetus has SMA.
- You can get pregnant using in vitro fertilization (IVF) . You can use your own eggs or sperm. You also can use donor eggs or sperm. Tests can be done on the embryo

before it is transferred to the uterus to see if it has SMA. This is called preimplantation genetic diagnosis .

- You can get pregnant using intrauterine insemination (IUI) . With this option, sperm from a donor who does not carry SMA would be used.
- You may choose not to get pregnant.
- You may choose to adopt a child.

Are my test results confidential?

Yes, but you may want to tell other family members if you find out that you are an SMA carrier. They may be at risk of being carriers themselves. There is no law that says you have to tell anyone. If you choose to tell family members, your obstetrician–gynecologist (ob-gyn) , genetic counselor , or other health care professional can give you advice about the best way to do this. Your test results cannot be shared without your consent.

I am concerned about discrimination based on genetic testing results. What should I know?

Many people are concerned about possible employment discrimination or denial of insurance coverage based on genetic testing results. The Genetic Information Nondiscrimination Act of 2008 (GINA) makes it illegal for health insurers to require genetic testing results or use results to make decisions about coverage, rates, or preexisting conditions. GINA also makes it illegal for employers to discriminate against employees or applicants because of genetic information. GINA does not apply to life insurance, long-term care insurance, or disability insurance.

Glossary

Amniocentesis: A procedure in which amniotic fluid and cells are taken from the uterus for testing. The procedure uses a needle to withdraw fluid and cells from the sac that holds the fetus.

Carrier: A person who shows no signs of a disorder but could pass the gene to his or her children.

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

Fetus: The stage of human development beyond 8 completed weeks after fertilization.

Gene: A segment of DNA that contains instructions for the development of a person's physical traits and control of the processes in the body. The gene is the basic unit of heredity and can be passed from parent to child.

Genetic Counselor: A health care professional with special training in genetics who can provide expert advice about genetic disorders and prenatal testing.

Genetic Disorder: A disorder caused by a change in genes or chromosomes.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Intrauterine Insemination (IUI): A procedure in which a man's semen is placed in a woman's vagina, cervix, or uterus.

Mutations: Changes in a gene that can be passed from parent to child.

Obstetrician–Gynecologist (Ob-Gyn): A doctor with special training and education in women's health.

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are done on the fertilized egg before it is transferred to the uterus.

Spinal Muscular Atrophy (SMA): An inherited disorder that causes wasting of the muscles and severe weakness. SMA is the leading genetic cause of death in infants.

If you have further questions, contact your ob-gyn.

Don't have an ob-gyn? Learn how to find a doctor near you.

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