

SMA carrier screening

Helping you make
an informed decision



SMA carrier screening Q&A

Q: If my healthcare provider doesn't suggest SMA carrier screening, does it mean I don't need to be tested?

A: While some healthcare providers offer it to their patients already, you may have to ask your healthcare provider about SMA carrier screening if they don't suggest it.

Q: My husband and I would never consider terminating a pregnancy. Why should we or couples who share our views consider SMA carrier screening?

A: While opposition to termination of a pregnancy is sometimes cited in arguments against genetic screening, it is important to realize that termination of a pregnancy is never the only option.

Q: Should I have SMA carrier screening even if my other children are not affected?

A: This is a personal decision that only you can answer and that may be influenced by several factors such as insurance, familiarity with the disease, personal opinions and beliefs regarding termination, and willingness to pursue other options if the test result is positive.

Q: Is SMA carrier screening harmful to me or my child if I'm pregnant while getting tested?

A: No, it is a blood test on the mother and should have no effect on the child.

Talk to your healthcare provider

It is important to discuss SMA carrier screening with your healthcare provider. To facilitate your conversation, here are some discussion points that may reflect your situation. SMA carrier screening puts you in control, helps you prepare, and gives you more options.

I already have a normal, healthy child or children

You and your spouse may still be SMA carriers even if you already have a healthy child or children. If you and your partner are both SMA carriers, there is a one in four chance with each pregnancy that you will have a baby with SMA.

I don't have a family history of SMA

You are at greater risk if someone in your family is affected by SMA. But even if you don't have a family history, you can still be a carrier of SMA and pass it on to your baby.

I'm under 40 years of age

Unlike some genetic disorders, such as Down syndrome, age has nothing to do with whether you could have a baby with SMA. Couples of all ages can be SMA carriers and have a baby with SMA.

Carrier screening won't change the outcome of an affected baby

If you know in advance about SMA, it can make a big difference in your baby's quality of life. It gives you time to prepare emotionally and financially. You will have more time to learn as much as you can about the disease and make arrangements for healthcare services. This will maximize your family's time together.

I'm not sure whether my insurance covers SMA screening

Insurance plans differ in their coverage. Check with your insurance provider to see if they cover SMA carrier screening and to what extent.

Additional resources

There are many resources available to learn more about SMA carrier screening. You can also get help if you and your partner are carriers or are expecting a child with SMA.

www.curesma.org

SMA is the number one genetic cause of death for infants. Cure SMA is dedicated to the treatment and cure of SMA. They fund groundbreaking research and provide families the support they need.

www.smafoundation.org

The mission of the Spinal Muscular Atrophy Foundation is to accelerate the development of a treatment for SMA.

www.fightsma.org

FightSMA is a volunteer, non-profit organization lead by parents. Its mission is to strategically accelerate research to treat or cure SMA.

What is SMA?

Spinal muscular atrophy (SMA) is a severe genetic disorder that may be fatal. Muscles involved in breathing, eating, or movement grow weaker until they waste away and die. SMA is the leading cause of death in early childhood.

- About one in every 40 people carries a mutated SMN1 gene, which can cause SMA.
- About one in every 6,000 - 10,000 babies is born with SMA every year.

There are three types of SMA

- The most common form, type I, which affects about 70% of patients, is the most severe. Children with type 1 SMA usually die from respiratory failure before the age of two.
- Children with type II SMA may be able to sit unaided, but cannot stand or walk unaided. These children typically live past age four.
- Children with type III SMA are able to walk unaided and have a normal lifespan, although they face many challenges.

Both parents must carry the gene

- A child can only have the disease if both parents carry the mutated SMN1 gene.
- When both partners are carriers, there is a 25% (one in four) chance with each pregnancy of having a child with SMA. (Of course, this means that there is a 75% chance that each pregnancy will not result in a child with SMA.)

Screening is simple and easy

- If you are planning to have a baby, a simple blood test can determine if you carry the gene for SMA. It is important that couples receive counseling and information. The screening should be voluntary and confidential.



**For more information
about SMA carrier
screening, speak to your
healthcare provider.**

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