What is SMA?

Spinal muscular atrophy (SMA) is a severe genetic disorder that causes progressive weakness of the muscles involved in breathing, eating, and movement. SMA affects one in 10,000 liveborn children and is the leading genetic cause of early childhood death, usually from respiratory failure. Although there is no cure for SMA, there are medications that have proven effective in treating SMA when started in infancy.

There are 3 types of SMA

- Type 1 SMA is the most common and severe form, affecting about 70% of patients. Children with type 1 SMA usually show signs of SMA before 6 months of age and often do not survive beyond 2 years of age
- Type II SMA is less severe than Type 1 with symptoms beginning after 6 months of age. Affected children may be able to sit unaided, but usually are unable to stand or walk without assistance. Most live into adulthood, depending on degree of breathing problems
- Type III SMA is more mild with symptoms beginning after 18 months of age. Affected children are usually able to walk and are expected to have a normal lifespan

Both parents must carry a gene variation in *SMN1*

- A child can have the disease only if both parents carry a change in the *SMN1* gene. This change is usually a missing portion (deletion) of the *SMN1* gene. Some carriers have 2 normal copies of the *SMN1* gene on 1 chromosome and a deleted *SMN1* gene on the other chromosome; this is called a silent carrier
- About 1 in 40 to 1 in 60 people are carriers for SMA. Carriers are not expected to have symptoms of SMA
- When both partners are SMA carriers, there is a 25% (1 in 4) chance with each pregnancy of having a child with SMA. 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 whether you are an SMA carrier. It is important that couples receive counseling and information. The screening should be voluntary and confidential
- Screening can identify most, but not all, SMA carriers. In addition to looking for the gene deletion, Quest Diagnostics also looks for a marker (called a single nucleotide polymorphism or SNP) that increases the chance the person is a silent carrier

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

Models used for illustrative purposes.

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SMA carrier screening

Helping you make an **informed decision**

For patients

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 many 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: It is important to realize that termination of a pregnancy is never the only option. For example, knowing an unborn baby may be affected with SMA might enable treatments to begin soon after birth for optimal effect.

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 1 in 4 chance with each pregnancy that you will have a baby with SMA.

I don't have a family history of SMA

Even if you don't have a family history, you can still be a carrier of SMA and pass it on to your child.

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, including consideration of newer medications that typically begin in infancy.

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.

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.

SMAFoundation.org

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

FightSMA.org

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

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