



Spinal Muscular Atrophy

DOCTOR DISCUSSION GUIDE FOR SMA CARRIER TESTING



Whether you are planning a pregnancy or are already pregnant, it is important to discuss SMA carrier testing with your doctor,

Why? Because even though the odds are that your baby will be healthy, you deserve to know in advance whether he or she might be born with SMA.

A simple blood test done in your doctor's office can tell you if you and your spouse carry the abnormal gene that causes SMA.* If the answer is no, you can be reassured that your baby is highly unlikely to have this incurable disorder. If the answer is yes, you will have numerous reproductive options [link] to consider with your genetic counselor.

Many doctors know about SMA and the benefits of SMA carrier testing and will recommend that you be tested before conception or early in pregnancy. But some doctors are not yet aware of the very high number of people who are SMA carriers (as many as one in every 40) or the benefits of SMA carrier testing, so you may want to bring this guide to your doctor and discuss if SMA Carrier testing is right for you.

To facilitate your conversation with your doctor, here are some discussion points about SMA carrier testing that may reflect your situation. Feel free to print this out and bring with you on your next doctor visit. Getting SMA carrier testing can be empowering because it 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 spouse are both SMA carriers, there is a one in four chance with each pregnancy that you will have a baby with SMA. The only way to know for sure if you are an SMA carrier is to get tested.

It's your future. SMA carrier testing puts you in control, helps you prepare, and gives you more options.

I DON'T HAVE A FAMILY HISTORY OF SMA

Knowing that you have a family history of SMA (either a relative with SMA or a family member who is an SMA carrier) puts you at greater risk of being an SMA carrier. But even if you don't have a family history of SMA, you can still be an SMA carrier and pass along the abnormal gene that causes the disease to your baby.

Because a baby with SMA must inherit two abnormal genes (one from the mother and one from the father), there can be generations of unaffected babies, leading couples and their doctors to mistakenly believe they could not be SMA carriers.



Recent studies suggest that up to one in every 40 people are SMA carriers, which means there are as many as 7.5 million SMA carriers in the United States alone.

You may have a family history of SMA and not even know it. Even with the most complete family history, there may be gaps. For example, one of your parents may be adopted, you may have lost contact with family members or not know the health history of deceased family members, or a relative may have had a baby who died in early infancy of undiagnosed SMA.

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I'M UNDER 40 YEARS OF AGE

Unlike a genetic disease 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.

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MY DOCTOR HASN'T HEARD OF SMA, SO DOESN'T BELIEVE CARRIER TESTING IS NECESSARY

Usually babies with SMA are born apparently healthy and often have the highest possible APGAR scores.* Often it is only when the baby is about three to six months old and fails to meet major developmental milestones that SMA is suspected and diagnosed by the child's pediatrician or a pediatric neurologist. Because your obstetrician/gynecologist probably sees few, if any, babies with SMA, he or she may not be aware of SMA or know that a large percentage of people in the general population are SMA carriers.

* The Apgar score is a method to check an infant's condition at one minute and five minutes after birth. The infant is checked for: Activity and muscle tone, Pulse (heart rate), Grimace response (reflex irritability), Appearance (skin color), and Respiration (breathing rate and effort). Each category is given a score ranging from 0-2, and numbers are added up for a final score. A perfect Apgar score is 10.

I AM ALREADY IN MY SECOND TRIMESTER OF PREGNANCY

Carrier testing requires only a simple blood test, so you can be tested at any point during your pregnancy with absolutely no risk to your baby. If testing reveals that both you and your spouse are SMA carriers (which means your baby has a one in four chance of having SMA), you can decide then whether to pursue prenatal diagnosis to determine whether your baby could be affected by SMA and, with Athena's SMA diagnostic testing, how severe his or her disease may be as determined by SMN2 copy number. Athena can help refer you to a qualified genetic counselor.

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CARRIER TESTING WON'T CHANGE THE OUTCOME OF AN AFFECTED BABY

While carrier testing won't change an affected baby's outcome, it can make a world of difference to you and your baby's quality of life. Knowing in advance that your baby has SMA gives you time to prepare emotionally and financially, learn as much as you can about the disease, make arrangements for healthcare services and assistive devices that can improve your baby's quality of life, and make important decisions that will maximize your family's time together.

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I'M NOT SURE WHETHER MY INSURANCE COVERS SMA CARRIER TESTING

Insurance plans differ in their coverage, so you should check with your insurance provider to find out if and to what extent they cover SMA carrier testing. Many couples feel that paying some costs out-of-pocket for a test that will give them such important information is well worth it.

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Only Athena offers the Patient Protection Plan, which limits the amount of out-of-pocket expenses for commercially insured patients. This program is available in 42 states*. Financial assistance is also available through Athena Diagnostics.

*The Patient Protection Program is for patients who currently have commercial insurance, and limits the amount of out-of-pocket costs to 20% of the cost of the test. Due to state laws, this program is not available in 8 states (MA, TX, NJ, MD, RI, FL, DE, OH)

WHAT IS SMA?

SMA is a severe, often fatal, genetic disorder in which muscles involved in many vital functions, such as breathing, eating, and movement, become progressively weaker and ultimately waste away (atrophy) and die.

SMA is the leading genetic cause of death in early childhood. Each year, about one in 10,000 infants is born with SMA, many of whom will not live past their second birthday.

Regardless of their race or ethnicity, up to one in every 40 people—as many as 7.5 million people in the United States alone—carries the abnormal gene responsible for SMA and can pass it along to their children. A simple blood test can tell couples whether they are SMA carriers.

For ordering the SMA test, please visit www.athenadiagnostics.com/sma

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