



REPRODUCTIVE GENETICS
Department of Obstetrics & Gynecology

Spinal Muscular Atrophy (SMA) Carrier Screening Information

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a relatively common genetic disease that affects approximately 1 of every 10,000 babies born. Boys and girls have an equal chance to have SMA. It is a chronic disease that usually grows progressively more severe throughout a person's life. Symptoms present in early infancy, starting with muscle weakness and a failure to meet milestones; in addition, joint contractures, tongue fasciculations, and absent reflexes also occur. SMA *does not* affect intelligence. The life expectancy for a person with SMA can be quite shortened; those with the infantile type of SMA typically pass away within the first 2-3 years of life due to respiratory failure if they do not receive treatment.

How is SMA treated?

There is currently no complete prevention or cure for SMA. FDA- approved treatments are currently available; Nusinersen and Zolgensma. Check with your institution to see if these treatments are readily available. Other treatment options consist of slowing symptom onset and promoting independence and comfort as much as possible.

How does a person get SMA?

SMA is an autosomal recessive disorder resulting from deletions and/or mutations in the *SMN1* and/or *SMN2* genes. For a child to have SMA, the child has to get an *abnormal* SMA gene from *both parents*. The parents of a child with SMA each carry *one abnormal* SMA gene and *one normal* SMA gene. Carrier parents do *not* have SMA and are perfectly healthy. Each time two carriers have a baby there is a 1 in 4 (25%) chance that the baby will have SMA.

Could I be a SMA carrier?

About 1 in every 40-60 Americans is a carrier for SMA. Most people who are SMA carriers do *not* have a relative with SMA and do not know they are carriers. If you *do have a relative with SMA*, then your chance to be an SMA carrier is greater than those chances listed above, and you should discuss this with your doctor or genetic counselor. The American College of Medical Genetics (ACMG) has recommended that all individuals of childbearing age be offered the option of SMA carrier screening, with appropriate follow-up with a genetic counselor to discuss abnormal results.

How can I find out if I am a SMA carrier?

A DNA test is available to all women to tell them if they are carriers for SMA. It is a regular blood test and can be ordered by your doctor or genetic counselor. If it says that you are a SMA carrier, you *definitely* are. However if it does not detect that you are a SMA carrier, there is still a small chance that you could be--the test identifies *most*, but not all SMA carriers. Once you have had the SMA carrier test, you will not need to have it again in future pregnancies or as you get older since the result will not change.

What if I am found to be a SMA carrier?

If a woman is found to carry the SMA gene, she will be referred to see a genetic counselor that will explain the result and help arrange to have her partner tested. Remember, it takes *both parents* being carriers for a baby to be at risk for SMA. If a woman's partner is not found to be a carrier, the couple could still have a baby with SMA, but the chance *with that partner* is very small. If a woman who is a SMA carrier becomes pregnant with a *different partner*, the new partner would also need to be tested to see if he is a carrier. If the woman and her partner are both SMA carriers, they have a 1 in 4 chance with each pregnancy to have a child with SMA. They could choose to test the fetus for SMA during the pregnancy if they would like to, and would have the option to end the pregnancy or continue the pregnancy if the fetus was found to have SMA. If they choose not to have prenatal testing, the baby could be tested after birth. A genetic counselor can discuss test options in greater detail.

Will SMA Carrier Screening be covered by my insurance?

Most of the major insurance companies in Rochester cover this testing. However since insurance coverage may vary, you should call your insurance company directly to inquire about your coverage.

How do I find out more about SMA Carrier Screening?

If you would like more information about SMA carrier screening, discuss it with your doctor, or call the Reproductive Genetics office at (585)487-3480.