



INFORMATION ABOUT SPINAL MUSCULAR ATROPHY

Spinal muscular atrophy (SMA) is a genetic disease that causes degeneration of the nerve cells, called motor neurons, in the spinal cord. These neurons communicate with the voluntary muscles (the ones we can control). As the neurons are lost, the muscles weaken. This can affect walking, crawling, breathing, swallowing, and head and neck control.

There are many types of SMA, and some of them are fatal. Life expectancy depends on the type and how it affects breathing. There is no cure. Medicines and physical therapy help treat symptoms.

Source: The National Institutes of Health

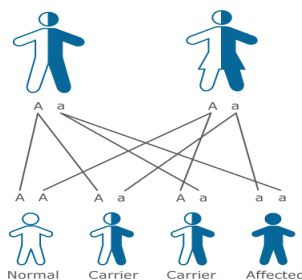
SPINAL MUSCULAR ATROPHY CARRIER SCREENING

What is carrier screening?

The American College of Obstetricians and Gynecologists recommends that couples planning a pregnancy, or those already pregnant, be informed about genetic carrier screening. Specific ethnic groups are known to have an increased risk for certain genetic conditions, and carrier screening is available to find out if a couple is at risk for having a child with one of these conditions. If both members of a couple are carriers for the same condition, there is a 25% chance that the pregnancy will inherit the condition. *It is important to note that these conditions often occur in families with no previous history of the condition.*

How are these conditions inherited?

The conditions included in most carrier screening tests are inherited in an “autosomal recessive” manner. This means that both parents must be “carriers” for a condition in order to be at risk for a pregnancy to be affected with that condition. If both parents are carriers for the same condition, the chance of having an affected pregnancy is 1 in 4 (or 25%) for *each* pregnancy, as illustrated in the diagram below.



Source: www.discern-genetics.org

A "carrier" is an individual with no symptoms of a particular disease, but who possesses both a normal gene (inherited from one parent) and a non-working gene (inherited from the other parent) for that condition.

A "non-carrier" is a individual who inherits two normal genes, one from each parent.

An "affected" person has inherited two non-working genes, or mutations, one from each parent.

Who should be screened?

Carrier screening is based on an individual's ethnicity and/or country of origin. With every ethnicity there are certain genetic disorders that are more common. Genetic carrier screening is available for many conditions that are more common in certain populations.

Genetic carrier screening for spinal muscular atrophy is offered to all individuals. Approximately 1/40 individuals is expected to be a carrier of spinal muscular atrophy.

When should carrier screening be performed?

It is strongly recommended that you and your partner undergo genetic carrier screening prior to pregnancy, or as early in pregnancy as possible.

What is the carrier screening process?

- In our office, genetic counseling is provided before carrier screening is ordered. This ensures that you are offered the most appropriate carrier screening options.
- Carrier screening is performed through a simple blood test; no preparation is necessary.
- Test results are provided to you and your physician within 2-3 weeks.
- If both partners are determined to be carriers for the same condition, additional genetic counseling will be available to discuss the nature of the specific disorder as well as prenatal testing options.

UNDERSTANDING YOUR RESULTS

What does a “screen negative” result mean?

A “screen negative” carrier screen result means that your chance of being a carrier for spinal muscular atrophy has been greatly reduced. A negative result significantly lowers, but does not completely eliminate, the chance of being a carrier. The percentage of carriers detected by our spinal muscular atrophy carrier screen is approximately 94%.

When your carrier screen is “screen negative”, we generally do not recommend that your partner pursue carrier screening, as the chance for an affected pregnancy is greatly reduced.

What does a “screen positive” result mean?

A “screen positive” carrier screen result means that you were found to be a carrier for spinal muscular atrophy. When the test determines that you are a carrier, the next step is for your partner to have carrier screening performed. Both parents must be carriers for the pregnancy to be at risk for spinal muscular atrophy. If testing determines that a couple is at risk, prenatal testing using chorionic villus sampling (CVS) or amniocentesis can be performed to determine whether the pregnancy has inherited the condition.

QUESTIONS?

If you have any questions concerning the above information, please do not hesitate to contact us at 312-981-4400.