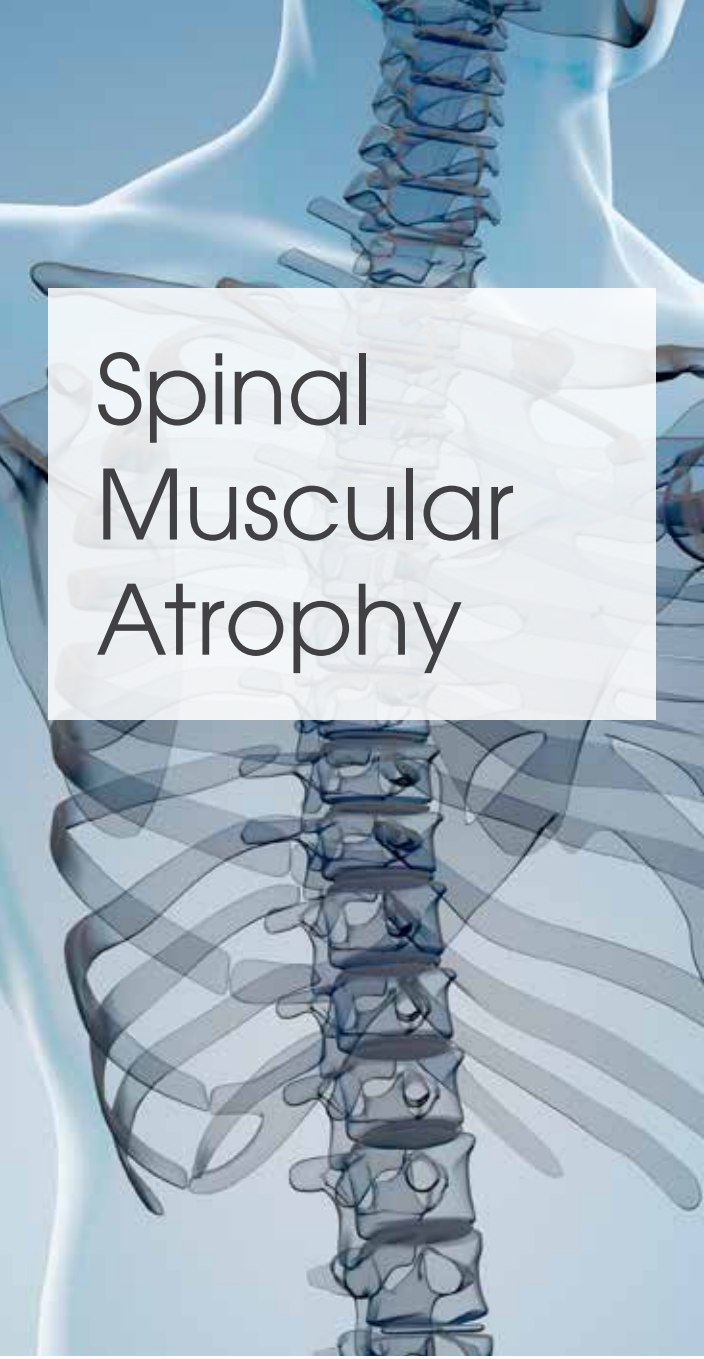

Spinal Muscular Atrophy

ARUP® LABORATORIES

Spinal muscular atrophy (SMA) is a genetic disorder that causes muscle weakness that may affect breathing, swallowing, and walking, especially in affected infants. With a single test, ARUP Laboratories can identify individuals who are either affected with SMA or are carriers of SMA .

Speak with a healthcare provider about your testing options.

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What is SMA?

SMA is a genetic disorder that primarily affects the nerves and muscles by damaging or destroying specific nerve cells in the spinal cord called motor neurons. This causes the muscles to become progressively weaker and smaller in those affected. The genes associated with most cases of SMA are called *SMN1* and *SMN2*.

There are several types of SMA. Symptoms can appear in infancy or not until adulthood, depending on the type of SMA. The most severe type causes breathing difficulties and serious muscle and nerve weakness, usually resulting in death before 2 years of age if not treated. Patients with other types can have more mild muscle weakness, and it may progress more slowly.

SMA is inherited from both parents, and approximately one in 6,000 to one in 10,000 children may be born with SMA.

Can my children have SMA even if no one else in my family does?

Yes. When this happens, it is most often because both parents are carriers of SMA, which means they each have one copy of the *SMN1* gene that does not work properly or is partially missing. Carriers of SMA usually do not have any symptoms and would not know they were carriers unless they had genetic testing. Two carriers can have a child who is affected with SMA.

Who can be a carrier of SMA?

Anyone can be a carrier of SMA. In the U.S. population, the overall chance of being an SMA carrier is one in 54.

What is the chance of having a baby with SMA if my partner and I are both carriers?

- There is a 25% chance that the baby will not have SMA and will not be carrier.
- There is a 50% chance that the baby will be a carrier of SMA, just like you and your partner.
- There is a 25% chance that the baby will have SMA.

What testing is available for SMA?

ARUP offers a first-tier diagnostic test for individuals suspected of having SMA, and this test is usually performed on a blood sample. The test includes copy number for the *SMN1* and *SMN2* genes, which means it detects how many copies of *SMN1* and *SMN2* a person has. Additionally, the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend carrier screening for SMA in all individuals who are pregnant or considering pregnancy.

If both parents are carriers of SMA, it is possible to do prenatal testing to find out if the pregnancy will be affected with SMA. Speak with your healthcare provider for help understanding your risks and testing options.

Is there a cure or treatment for SMA?

There is no cure for SMA. Treatment for SMA is available, depending on the specific type.

Disease-modifying treatments include medications that can slow the progression of the disease and improve muscle strength in patients with SMA.

In 2019, the U.S. Food and Drug Administration (FDA) approved gene replacement therapy for children younger than 2 years with SMA. This treatment also slows the disease progression and improves muscle strength in children with SMA.

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