

Supporting Rare Disease – SMA Awareness Month

Rare Disease Support

There are times when every hour is critical to a patient's health. At CuraScript SD, we ensure that every rare disease patient gets the critical medication they need within the hours they need it. We provide integrated solutions for the safe and expedited distribution of rare and orphan specialty pharmaceuticals. Essential therapies and customized business solutions come together to enhance efficiency while delivering pivotal care.

Spinal Muscular Atrophy Awareness

Spinal Muscular Atrophy (SMA) affects the motor nerve cells in the spinal cord, robbing patients of physical strength and impacting their ability to walk, eat, and breathe. It is one of the most common genetic causes of death among infants.

Each August, healthcare providers and SMA community members help raise awareness about this rare disease. In an effort to help raise public awareness about SMA, please note the following information about this rare disease.

SMA Cause & Symptoms

Spinal Muscular Atrophy is caused by an autosomal recessive gene, meaning that in most cases, both parents must carry the genetic mutation for a child to have the condition. Symptoms vary greatly. In more severe cases, they generally occur

in the first six months of life; in milder cases, symptoms may not be noticeable until 18 months or older. Symptoms may include:

- Muscle weakness & decreased muscle tone
- Limited mobility
- Breathing problems
- Problems eating & swallowing
- Delayed gross motor skills
- Spontaneous tongue movements
- Scoliosis (curvature of the spine)

Diagnosing SMA

If SMA is suspected, your healthcare provider may conduct the following tests to confirm a diagnosis:

- Genetic blood tests
- EMG to measure electrical activity of a muscle group(s)
- CPK test to rule out other neuromuscular diseases

Promising Treatment Options

The discovery of the genetic cause of SMA has led to the development of two new treatment options. These have shown promising results.

- SMA gene therapy is a one-time infusion that has been proven to improve motor neuron function and increase the likelihood of survival.
- A second treatment uses a synthetic material to help patients make more SMN protein needed to improve healthy motor neurons.

Did you know?

1 in 50 people carry a copy of the altered gene that can lead to SMA in a child.

Learn More

Click [here](#) for more information about how CuraScript SD supports rare disease healthcare providers and their patients.

References:

<https://www.curesma.org/sma-awareness-month/#:~:text=About%20SMA%20Awareness%20Month,and%20those%20impact%20by%20SMA>

<http://www.childrenshospital.org/conditions-and-treatments/conditions/s/spinal-muscular-atrophy-sma/diagnosis-and-treatment>