

SPINAL MUSCULAR ATROPHY

1 in 50 Americans are Carriers of SMA

SMA is the Leading Genetic Cause of Death in Infants

Spinal Muscular Atrophy (SMA) is a rare neuromuscular disease. SMA has generally been believed to affect as many as 10,000 to 25,000 children and adults in the United States, and therefore it is one of the most common rare diseases.

This resource will provide valuable information including, what Spinal Muscular Atrophy is, causes, symptoms and care options.

01 | What is Spinal Muscular Atrophy?

Spinal muscular atrophy (SMA) is a genetic disease affecting the part of the nervous system that controls voluntary muscle movement. SMA involves the loss of nerve cells called motor neurons in the spinal cord and is classified as a motor neuron disease.

SMA is a disease that robs people of physical strength by affecting the motor nerve cells in the spinal cord, taking away the ability to walk, eat, or breathe. It is the number one genetic cause of death for infants.

02 | Causes

The most common forms of SMA are caused by a defect (mutation) in the SMN1 gene on chromosome 5. Those with SMA have two SMN1 genes, one on each chromosome 5. Normally, SMN1 genes produce full-length and fully functional SMN protein. However, when the SMN1 gene has mutations, insufficient levels of SMN protein are produced.

03 | Symptoms

Symptoms of SMA vary greatly depending on SMA type. More severe forms of SMA affect more essential functions that are very apparent during the first 6 months of life and less severe forms of SMA have symptoms that are not noticed until later in life. Symptoms include:

- Muscle weakness and decreased muscle tone
- Weakness on the right and left sides of the body
- Leg immobility
- Limited mobility due to muscle weakness
- Lack of weight bearing functions
- Fragile bones and fractures
- Breathing problems especially in young children
- Weak tongue and swallowing difficulties
- Scoliosis

04 | Care Options

Currently there is no cure for SMA. Treatment consists of managing symptoms and preventing complications. Common care options include:

- Muscle relaxants – assist in reducing spasticity
- Physical therapy – assist in improving posture, preventing immobility and slow down muscle weakness and atrophy
- Proper nutrition – is essential in maintaining weight and strength

Additional information is available regarding SMA, for resources and support, visit

<https://www.mda.org/disease/spinal-muscular-atrophy>

Did You Know?

1 in 10,000 babies are born with SMA

References

<http://www.curesma.org/sma/about-sma/>

<https://www.mda.org/disease/spinal-muscular-atrophy/causes-inheritance>

<http://www.childrenshospital.org/conditions-and-treatments/conditions/spinal-muscular-atrophy-sma/symptoms-and-causes>