

SPINAL MUSCULAR ATROPHY (SMA) – A GENETIC MOTOR NEURON DISEASE

SMA Affects Approximately 1 in 10,000 Babies

Spinal Muscular Atrophy is the Leading Cause of Genetic Death for Infants

Spinal Muscular Atrophy (SMA) is a genetic disease caused by a mutation that leads to debilitating and sometimes fatal muscle weakness. There are four primary types of SMA. Please read the following information to learn more about the disease, its effects and what treatment options are available.

01 | Description and Types

SMA is a genetic neuromuscular disease resulting in muscle weakness and atrophy. It typically manifests early in life and is caused by defects in the Survival Motor Neuron 1 (SMN1) gene. There are four main types of SMA, which include the following:

- **Type 1: Symptoms present by 6 months.** This is the most severe form of the disease and present during infancy. Children are unable to sit up, crawl or walk, and basic respiration is a critical risk.
- **Type 2: Symptoms present by 18 months.** Respiratory symptoms may occur before 18 months. Head control, turning and rolling in the crib are not achieved, and a child affected by Type 2 SMA is unlikely to walk unassisted.
- **Type 3: Onset in childhood.** Many children are diagnosed by age 3, but diagnosis can occur between ages 18 months to early adolescence. Children with Type 3 SMA can be ambulatory but may lose the ability to walk later in life.
- **Type 4: Adult onset.** Diagnosed between ages 15 to 50, and symptoms include muscle wasting, weakness and tremors.

02 | Treatment

While there is currently no cure for SMA, there are treatment options for specific symptoms:

- **Respiratory muscle weakness:** Noninvasive ventilation can be delivered under pressure through a mask or mouthpiece. Negative-pressure ventilation systems, which create an intermittent vacuum around the chest to help the lungs expand contract can also

be used. In some cases, a tracheostomy may be recommended in early childhood.

- **Swallowing muscle weakness:** Infants with swallowing or sucking weakness can be fed with a gastrostomy tube.
- **Back muscle weakness:** Bracing has been found to be a good source of support in children with SMA, especially in preventing scoliosis. Spinal fusion may also become necessary.
- **Exercise, physical therapy and occupational therapy:** Most physicians advise doing as much physical activity as is comfortable, to protect the joints from stiffness, preserve range of motion, and maintain circulation.

03 | The Search for a Cure

The genetics of SMA provides a unique prospect for therapeutic development. While patients lack the functional SMN1 gene, they carry a backup gene, SMN2, which also makes SMN protein, but at a much lower efficiency. This provides hope for the development SMA therapeutics, and the majority of drug development efforts are focused on increasing SMN protein production from this gene.

For more information on SMA and new developments, please visit: <http://www.curesma.org/sma/about-sma/>

*Did You Know?
1 in every 50 Americans is a
genetic carrier of SMA*

References

<http://www.mayoclinic.org/diseases-conditions/heart-disease/in-depth/heart-disease-prevention/art-20046502>

<http://mylifecheck.heart.org/Multitab.aspx?NavID=3&CultureCode=en-US>