

SPINAL MUSCULAR ATROPHY

Spinal muscular atrophy is present in approximately 1 in 10,000 live births

01 | Introduction

Spinal Muscular Atrophy (SMA) is a disease that affects both the nervous system and skeletal muscle. SMA is most often caused by genetic mutations in the *SMN1* gene. This gene codes for a motor neuron protein, SMN, which helps motor neurons to properly perform their functions. In cases of SMA, because the motor neurons are not sufficiently supported by the SMN proteins, they cannot send signals to the muscles. Over time, this causes the muscles to become weaker and shorten (atrophy) as they are not being stimulated by their corresponding neurons.

02 | Signs and Symptoms

Many people with SMA have the following symptoms that result from the weakened muscles:

- Scoliosis
- Respiratory issues
- Upper and lower limb weakness
- Decreased motor function

03 | Types

There are several different types of SMA which are classified based upon severity and onset.

- **Type 0:** The most severe type of SMA. Infants born with this type often do not survive past 6 months.
- **Type 1:** Disease onset at birth or by 6 months. Most children will not survive past 2 years of age.
- **Type 2:** Disease onset between 3 and 15 months. Some children will survive into adulthood.
- **Type 3:** A milder form of SMA that appears between 18 months and adulthood.
- **Type 4:** The mildest form of SMA. Disease onset occurs around age 30.

03 | Treatments

There are two main treatments that are used for patients with SMA. The first is a single dose gene therapy that replaces the faulty *SMN1* gene with a working one. The other is an injection medication that is administered multiple times per year. Although neither treatments are considered cures for SMA, they both have shown improvements in the lives of those who have received them.

For more information on spinal muscular atrophy and supportive resources, please visit: www.mda.org.

Did You Know?

Mutations in the SMN1 gene are present in 95% of people with spinal muscular atrophy

References

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

<https://www.mayoclinic.org/what-is-spinal-muscular-atrophy/art-20491890>