

LEARNING ABOUT SPINAL MUSCULAR ATROPHY

Spinal Muscular Atrophy (SMA) is diagnosed in 1 in 100,000 people.

Spinal Muscular Atrophy (SMA) is a rare genetic condition where nerve cells in the spinal cord that control the muscles are damaged. Please use this guide as a resource for knowledge and understanding of SMA.

01 | Cause

SMA is most often caused by a mutation in a gene on chromosome 5. Both parents must carry a defective gene in order for a child to be affected. There are several variants of the disease and the severity of the disease can range from mild impairment to early childhood morbidity.

02 | Symptoms

SMA is typically diagnosed when a child fails to meet normal physical milestones. In more severe forms of SMA in infancy, an infant will present as a “floppy baby.” The infant will have little to no motor strength or control. It can be difficult to feed from a bottle or breastfeed and the child’s hands and arms become weaker before other muscles are affected.

03 | Diagnosis

Spinal Muscular Atrophy is usually diagnosed by genetic testing. A blood test is performed to check for the presence of the SMN1 gene. In about 95% of those with SMN-related SMA, this gene will be missing. In the other 5%, the gene will appear mutated. Other tests such as an EMG or muscle biopsy may also be performed.

04 | Treatment & Care

Although there is no known cure for SMA, new gene therapy treatments have been developed recently that help to give the body instructions for making a protein that helps with controlling muscle movement. These therapy options, along with treatment to assist with breathing, feeding, physical and occupational therapy can help with symptoms.

Treatment of a child with Spinal Muscular Atrophy requires a team of doctors and therapists. Support groups can be a great resource for parents and caretakers of children with SMA.

For more information on SMA and to find support resources, please visit: <https://www.smafoundation.org/>

Did You Know?

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

References

<https://simedhealth.com/article/spinal-muscular-atrophy-information>

<https://my.clevelandclinic.org/health/diseases/14505-spinal-muscular-atrophy-sma>

<https://www.webmd.com/a-to-z-guides/spinal-muscular-atrophy#5>