



SPINAL MUSCULAR ATROPHY

MEDICAL FOCUS

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Spinal Muscular Atrophy, or SMA, is a progressive motor neuron disease resulting in muscle weakness. Motor neurons in the spinal cord that typically send signals to muscles deteriorate and as the amount of signals decrease, the muscles atrophy. SMA is an autosomal recessive genetic disease, meaning that two gene flaws must occur – one from each parent. The parent, in this case, is a carrier but has no symptoms. One in 50 people are carriers of this disease. One in 6,000 to 1 in 10,000 children are born with this disease, which is the primary genetic cause of death in infants and toddlers. Between 10,000 and 25,000 children and adults in the United States have a form of SMA.

The most common form of SMA is caused by a deficiency of the SMN motor neuron protein. SMN protein, or “survival of motor neuron,” is required for normal motor neuron function. Genetic flaws on each chromosome 5 in a gene called SMN1 lead to this protein deficiency. The protein made by normal SMN1 genes is full-length and functional. Another gene, SMN2, is also present and not flawed. Most of the protein made by SMN2 genes is short and not functional, but some is full-length and functional. As a result, SMN2 genes can partially make up for the loss of SMN. The number of SMN2 genes vary from person to person and the more SMN2 gene copies a person has, the later the onset of the disease will be. Genetic testing can tell how many SMN2 genes are present and helps to predict the course of the disease.

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Current research is working on means to increase production of SMN protein, as well as strategies to help motor neurons survive.

Symptoms include muscle weakness in the voluntary muscles, with most weakness occurring proximally, or close to the body. Sensory and cognitive skills remain intact.

Although the most common form of SMA is categorized into 4 types (I – IV), some physicians believe this is more of a continuum. In general, the later the symptoms begin and the more SMN protein is present, the milder the course of the disease will be.

Type I SMA, also called Werdnig–Hoffman disease, presents between birth and age 6 months. These babies do not reach the developmental milestone of sitting. Two copies of the SMN2 gene are usually present. Over half of new SMA cases are type 1.

Type 2 SMA presents between the ages of 7 and 18 months, before the child can stand or walk independently. At least three SMN2 genes are usually present.

Type 3 SMA presents after 18 months and standing and walking are typically possible, though walking aids may be required. Usually four to eight SMN2 genes are present.

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RESOURCES AND REFERENCES:

MUSCULAR DYSTROPHY ASSOCIATION, SPINAL MUSCULAR ATROPHY
[HTTPS://WWW.MDA.ORG/DISEASE/SPINAL-MUSCULAR-ATROPHY](https://www.mda.org/disease/spinal-muscular-atrophy)

NATIONAL INSTITUTES OF HEALTH U.S. NATIONAL LIBRARY OF MEDICINE, SPINAL MUSCULAR ATROPHY
[HTTPS://GHR.NLM.NIH.GOV/CONDITION/SPINAL-MUSCULAR-ATROPHY](https://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy)

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[HTTP://WWW.SMAFOUNDATION.ORG/](http://www.smafoundation.org/)

