

What is spinal muscular atrophy?

Spinal muscular atrophy (SMA) is an inherited disease with variable severity and age at onset. Individuals with spinal muscular atrophy have a reduction in the normal amount of survival motor neuron protein,^{1,2} which leads to the destruction of nerves responsible for controlling voluntary muscle movement.¹ Intelligence is not affected.² Spinal muscular atrophy is also known as congenital axonal neuropathy, arthrogryposis multiplex congenita (prenatal SMA), Werdnig-Hoffman disease (SMA type I), Dubowitz disease (SMA type II) and Kugelberg-Welander disease (SMA type III).²

What are the symptoms of spinal muscular atrophy and what treatment is available?

Spinal muscular atrophy is characterized by progressive degeneration of the lower motor neurons in the spinal cord and brain stem, leading to muscle weakness and, in its most common form, respiratory failure by age two.² Muscles responsible for crawling, walking, swallowing, and head and neck control are the most severely affected.¹ Complications may include poor weight gain, sleep difficulties, pneumonia, scoliosis (curvature of the spine), and joint deformities.² Ages at onset and disease symptoms are variable, ranging from a severe prenatal form to a milder adult form primarily characterized by muscle weakness.² It is estimated that 60% to 70% of individuals who are diagnosed with spinal muscular atrophy are severely affected.³

There is no cure for spinal muscular atrophy. Treatment may include supportive care and surgical intervention as needed.²

How is spinal muscular atrophy inherited?

Spinal muscular atrophy is an autosomal recessive disease caused by mutations in the *SMN1* gene.¹ Individuals who inherit one copy of *SMN1* are carriers and are not expected to have related health problems. Individuals who inherit two or more copies of *SMN1* have a reduced carrier risk. Approximately 95% of affected individuals have 0 copies of *SMN1*.⁴

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for spinal muscular atrophy?

Spinal muscular atrophy is the most common inherited cause of early childhood mortality and can occur in individuals of all races and ethnicities.

Select carrier frequencies⁴

Population	Carrier Frequency
African American	1 in 72
Ashkenazi Jewish	1 in 67
Asian	1 in 59
Asian Indian	1 in 52
Caucasian	1 in 47
Hispanic	1 in 68

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Muscular Dystrophy Association: <https://www.mda.org/disease/spinal-muscular-atrophy/overview>

Spinal Muscular Atrophy Foundation: <http://www.smafoundation.org/>

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy>

Claire Altman Heine Foundation: www.preventsma.org

References

1. Spinal muscular atrophy. *Genetics Home Reference*. Available at <http://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy>. Accessed January 20, 2016.
2. Prior T and Russman B. Spinal Muscular Atrophy. *GeneReviews*. Available at <http://www.ncbi.nlm.nih.gov/books/NBK1352/>. Accessed January 20, 2016.
3. Meldrum C *et al*. Spinal muscular atrophy genetic counseling access and genetic knowledge: Parents' Perspectives. *J Child Neurol*. 2007 22:1019-1026.
4. Sugarman EA *et al*. Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. *Eur J Hum Genet* 2012; 20: 27-32.