Genetics Uncoded:

NxGen MDx

Facts about
Spinal Muscular Atrophy

What Your Test Results Mean

Carriers show no symptoms of spinal muscular atrophy and are not at risk to develop the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Spinal Muscular Atrophy Explained

Spinal muscular atrophy is an inherited neuromuscular disorder characterized by progressive muscle weakness caused by spinal cord and brainstem motor neuron degeneration. Individuals with spinal muscular atrophy do not produce enough of one of the motor neuron proteins, SMN, needed for proper motor neuron function.

There are several types of spinal muscular atrophy including severe, intermediate, mild, and adult. The clinical spectrum ranges from early infant death in severe type I to normal adult life with only mild weakness in adult type IV. Severe type I spinal muscular atrophy presents with muscle weakness by six months of age and inability to sit independently. Poor weight gain, sleep difficulties, pneumonia, scoliosis, and joint contractures are common. In intermediate type II, and mild type III, some children sit but never walk, whereas others show delayed walking but may be able to maintain walking until adult years. Pulmonary disease is a major complication of the disease in addition to gastrointestinal dysfunction and limited motor function of the trunk and extremities. In adult type IV, onset occurs in the second or third decade of life. Motor impairment is mild without respiratory or gastrointestinal problems.

Treatment of individuals with spinal muscular atrophy is supportive and typically provided by a team of specialists including pulmonology, gastroenterology, orthopedics, and others.

How the Genetics Work

The clinical features of spinal muscular atrophy can be explained by a deletion in both copies of the *SMN1* gene. In general, individuals have two copies of the *SMN1* gene. Carriers of spinal muscular atrophy have a deletion in one copy of the *SMN1* gene while individuals with spinal muscular atrophy have deletions in both copies of their genes. In addition, it is suspected that the number of copies of the *SMN2* gene may play a role in the severity of disease. A higher copy number of *SMN2* has been associated with milder disease; however, predicting the severity of disease using *SMN2* copy number is not currently recommended as phenotype can be variable.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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