NxGen MDx

Spinal Muscular Atrophy

What Your Results Mean

Test results indicate that you are a carrier of spinal muscular atrophy. Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for spinal muscular atrophy each of your children has a 1 in 4 (25%) chance to have the condition.

In approximately 2% of cases, a person with spinal muscular atrophy inherits an *SMN1* gene variant from one parent and acquires a new variant in the other copy of the gene that occurs during the formation of reproductive cells (eggs or sperm) or in early embryonic development. In these cases, only one parent is a carrier of the *SMN1* gene variant.

Spinal Muscular Atrophy Explained

What is Spinal Muscular Atrophy?

Spinal muscular atrophy (SMA) 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 five main subtypes of spinal muscular atrophy, each described below. It is not always possible to predict which type of SMA a child could have based on the genetic mutation they inherit.

Type 1 *SMN1*-linked SMA (also called Werdnig-Hoffman disease) is the most severe type of SMA. Babies born with SMA type 1 show symptoms of the condition within the first six months of life, experience significant difficulty with breathing and swallowing, and are unable to sit without support. Individuals with type 2 *SMN1*-linked SMA (also called Dubowitz disease) typically develop symptoms between six and twelve months of age. Although they do learn to sit up without support, children with SMA type 2 cannot stand or walk without help. Type 3 *SMN1*-linked SMA is called Kugelberg-Welander disease or juvenile type SMA and it is a milder form of the condition. These individuals typically have symptoms in early childhood, but they often learn to stand and walk without support and can maintain this ability into their thirties or forties. Type 4 *SMN1*-linked SMA is the mildest form of the condition, with symptoms typically appearing during adulthood. These individuals can experience some muscle weakness, tremors, and twitching.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
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Prognosis

The prognosis for individuals with SMA depends on the severity of the symptoms. Children with SMA type 1 typically have a poor prognosis, as affected infants experience significant difficulties breathing and swallowing, and typically die around two years of age. There is some evidence that children now live slightly longer with improved respiratory care and nutrition. The prognosis for individuals with SMA type 2 is better than that of children with type 1, as more than two thirds of affected individuals can live into their twenties. The prognosis for individuals with SMA Type 3 is good, though they can lose the ability to walk in their thirties and forties and then require mobility aids. The prognosis for individuals with SMA Type 4 is favorable, as symptoms appear in adulthood and their mobility remains relatively intact.

Treatment

Although there is no cure for SMA, the symptoms of the condition can be managed to maximize an individual's quality of life. For children with SMA type 1, treatments focus around making the child comfortable, including maximizing their breathing capacity for as long as possible and ensuring that they receive appropriate nutrition despite feeding and swallowing difficulties. Scoliosis can be a significant issue for individuals with SMA types 2 and 3, so these individuals should receive appropriate orthopedic care. Individuals with SMA may also be prone to disordered sleeping, including snoring and sleep apnea, and they should receive appropriate treatment. In general, individuals with SMA should be evaluated at least every six months to assess their general health.

Additionally, a medication is now available that has been shown to improve motor development in infants and children with the condition. This medication, known as nusinersen (market name Spinraza™), has been approved in the United States for use in pediatric and adult patients with SMA.

Resources

Claire Altman Heine Foundation
http://www.clairealtmanheinefoundation.org/
Families of Spinal Muscular Atrophy (Families of SMA)
http://curesma.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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