NxGen MDx

Sialic Acid Storage Disorders

What Your Results Mean

Test results indicate that you are a carrier of sialic acid storage disorders. 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.

Recommended Next Steps

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

Sialic Acid Storage Disorders Explained

What are Sialic Acid Storage Disorders?

Sialic acid storage disorders are inherited disorders that affect an individual's nervous system. They cause slow, progressive decline in motor and mental skills. The disorder is generally classified into one of three forms: infantile free sialic acid storage disease, Salla disease, and intermediate severe Salla disease.

Infantile free sialic acid storage disease (ISSD) is the most severe form of this disorder. Babies with this condition have severe developmental delay, weak muscle tone, and failure to gain weight and grow at the expected rate. They may have coarse facial features, seizures, bone malformations, an enlarged liver and spleen, and an enlarged heart. The abdomen may be swollen due to the enlarged organs and an abnormal buildup of fluid in the abdominal cavity (ascites). Affected infants may have a condition called hydrops fetalis in which excess fluid accumulates in the body before birth. Children with this severe form of the condition usually live only into early childhood.

Salla disease is a less severe form of sialic acid storage disease. Babies with Salla disease usually begin exhibiting weak muscle tone during the first year of life and go on to experience progressive neurological problems. Signs and symptoms of Salla disease include intellectual disability and developmental delay, seizures, problems with movement and balance (ataxia), abnormal tensing of the muscles (spasticity), and involuntary slow, sinuous movements of the limbs. Individuals with Salla disease usually survive into adulthood.

People with intermediate severe Salla disease have signs and symptoms that fall between those of ISSD and Salla disease in severity.

Prognosis

The prognosis for individuals with one of the sialic acid storage diseases can vary depending on the type and severity of the individual's symptoms. Children with the ISSD form of the condition usually live only into early childhood. While the lifespan for people with Salla disease is typically not significantly shortened, these individuals are also typically quite disabled and most likely need lifelong care, as only one third of affected children learn to walk and speech can be poor. Few cases of intermediate severe Salla disease are known, making it difficult to describe the prognosis for affected individuals.

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Treatment

There is no cure available for any of the sialic acid storage diseases, but treatments do exist to manage some of the symptoms and improve individuals' quality of life. In general, people with any type of sialic acid storage disease benefit from therapies aimed at optimizing their mobility and motor function (physical and occupational therapy) as well as their communication abilities (speech therapy). Those individuals who reach school age should attend schools with adequate special education programs. Individuals should also receive the standard medical treatments to manage their seizures and any other complications.

Resources

Hide & Seek Foundation for Lysosomal Disease Research
https://hideandseek.org

National Society of Genetic Counselors
https://www.nsgc.org/

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