Canavan Disease

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Canavan disease. Carriers are not expected to show symptoms of Canavan disease. You and your partner would both have to be carriers of Canavan disease for there to be an increased chance to have a child with symptoms. This is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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 or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for Canavan disease, each of your children has a 1 in 4 (25%) chance to have the condition.

Canavan Disease Explained

What is Canavan Disease?

Canavan disease is an inherited metabolic disorder characterized by the inability of the nerve cells to receive and send messages. There are two forms of Canavan disease, neonatal/infantile and mild/juvenile. In the more common form, neonatal/infantile Canavan, infants appear normal during the first couple months of life, but symptoms begin to develop by five months of age. Infants are unable to develop motor skills or meet developmental milestones, such as holding their head up, within the first year of life. Additional symptoms include sleep disturbance, weak muscle tones, a large head, seizures, and feeding difficulties.

The less common form of Canavan disease is the mild/juvenile form. These individuals have mild speech delays and delayed motor skills typically apparent in childhood. Usually these delays are so mild that they may not be readily recognized as being caused by Canavan disease.

Prognosis

Prognosis is considered unfavorable for the neonatal/infantile form of Canavan disease. Life expectancy is variable, ranging from early childhood to teenage years. Individuals with the mild or juvenile form do not appear to have a shortened lifespan.

Treatment

Treatment of individuals with Canavan disease is supportive as there is no cure. Affected individuals benefit from treatment focusing on maintaining nutrition and hydration, managing infectious diseases, and seizures.

Resources
Canavan Foundation
http://www.canavanfoundation.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/canavan-disease/
National Society of Genetic Counselors
https://www.nsgc.org/

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