NxGen MDx

Krabbe Disease

What Your Results Mean

Test results indicate that you are a carrier of Krabbe disease, sometimes referred to as globoid cell leukodystrophy. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of Krabbe 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 or donor are carriers for Krabbe disease, each of your children has a 1 in 4 (25%) chance to have the condition.

Krabbe Disease Explained

What is Krabbe Disease?

Krabbe disease is an inherited metabolic disorder that affects nerve cells of the central nervous system. Deficient levels of the enzyme galactosylceramide beta-galactosidase lead to a build-up of substances that damage the myelin sheath of nerve cells. Krabbe disease can be diagnosed in infancy or later in life. Infantile Krabbe disease is generally fatal before age two. Individuals with juvenile or adult-onset Krabbe disease generally have a milder course of the disease and live significantly longer. Symptoms include muscle weakness, irritability, feeding difficulty, fevers without sign of infection, and slowed mental and physical development. Over time, affected individuals have difficulty moving, swallowing, chewing, and breathing and develop hearing and vision loss and seizures. At this time, it is difficult to use genetic testing results to predict when onset of symptoms will occur.

Prognosis

Prognosis is generally poor. Infantile Krabbe disease is typically fatal before age two. Individuals with later onset usually die within 2-7 years after the onset of symptoms.

Treatment

Treatment is primarily supportive. Bone marrow and stem cell transplantation have been used with variable results in treating disease.

Resources

United Leukodystrophy Foundation
https://ulf.org/krabbe-disease/

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/krabbe-disease

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

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