Genetics Uncoded:

Facts about Krabbe Disease

What Your Test Results Mean

Carriers typically show no symptoms of Krabbe disease; however, carriers are at an increased risk of having a child with Krabbe disease. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Krabbe Disease Explained

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. At this time, it is difficult to use genetic testing results to predict when onset of symptoms will occur. Bone marrow and stem transplantation have been used with variable results in treating disease.

How the Genetics Work

Krabbe disease is an autosomal recessive disorder caused by pathogenic variants in the *GALC* gene. In general, individuals have two copies of the *GALC* gene. Carriers of Krabbe disease have variants in one copy of the *GALC* gene, while individuals with Krabbe disease have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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