Genetics Uncoded:
FACTS ABOUT KRABBE DISEASE

AKA | Diffuse Globoid Body Sclerosis, Galactosylceramide Deficiency Disease, Galactosylceramide Lipidosis, Galactosylcerubrosidase deficiency, Galactosylsphingosine Lipidosis, GALC deficiency, GCL, GLD, Psychosine Lipidosis

What Your Test Results Mean
Testing results indicate that you are a carrier of Krabbe disease. Carriers typically show no symptoms of Krabbe disease; however, carriers are at an increased risk of having a child with Krabbe disease. Risk for the 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.

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 2. 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 mutations in the GALC gene. In general, individuals have two copies of the GALC gene. Carriers of Krabbe disease have mutations in one copy of the GALC gene, while individuals with Krabbe disease have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps
Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?
Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.