Phosphoglycerate Dehydrogenase Deficiency/Neu-Laxova Syndrome

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of phosphoglycerate dehydrogenase deficiency. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of phosphoglycerate dehydrogenase deficiency 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 phosphoglycerate dehydrogenase deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Phosphoglycerate Dehydrogenase Deficiency Explained

What is Phosphoglycerate Dehydrogenase Deficiency?

Phosphoglycerate dehydrogenase deficiency is an inherited condition that prevents the body from synthesizing the amino acid serine. Serine is very important in the brain in the formation of neurotransmitters, neuron cell membranes, and myelin sheath to protect the neurons. The lack of serine causes serious neurological symptoms. Individuals with the more severe infantile onset form may have small head size identifiable by ultrasound during pregnancy. They also typically have recurrent seizures and psychomotor delays within the first year of life. In juvenile onset phosphoglycerate dehydrogenase deficiency, mild developmental delay and seizures are common. Administration of serine during pregnancy has been used to manage the infantile onset form and oral serine has proven to prevent seizures in the juvenile onset form.

Prognosis

The prognosis for phosphoglycerate dehydrogenase deficiency is generally poor. Certain treatments can assist with seizures, but the other symptoms remain and are somewhat progressive.

Treatment

Amino acid therapy has shown a favorable response in the reduction of seizure activity. Psychomotor development due to high doses of L-serine was only noted in one patient. Prenatal serine dosage was shown to increase head circumference.

Resources

Genetic Home Reference
https://ghr.nlm.nih.gov/condition/phosphoglycerate-dehydrogenase-deficiency#statistics
National Society of Genetic Counselors
https://www.nsgc.org/

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