Congenital Disorder of Glycosylation, *PMM2*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of congenital disorder of glycosylation, *PMM2*-related (CDG-*PMM2*). Carriers are not expected to show symptoms of CDG-*PMM2*. You and your partner or donor would both have to be carriers of CDG-*PMM2* 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 CDG-*PMM2*, each of your children has up to a 1 in 3 (33%) chance to have the condition. This risk is higher than the 1 in 4 (25%) chance expected for other autosomal recessive conditions.

Congenital Disorder of Glycosylation, *PMM2*-Related Explained

What is Congenital Disorder of Glycosylation, *PMM2*-Related?

Congenital disorder of glycosylation, *PMM2*-related (CDG-*PMM2*) is an inherited metabolic condition that affects multiple organ systems of the body. Symptoms of CDG-*PMM2* usually start shortly after birth. Infants can experience global brain dysfunction, poor muscle tone, abnormal eye movement, and psychomotor delays. More severe cases of CDG-*PMM2* present with hydrops fetalis, a fatal condition in which excess fluid builds up in the body before birth. Affected individuals with CDG-*PMM2* who survive infancy may have intellectual disability, may not be able to walk independently, and may experience stroke-like episodes involving temporary paralysis.

Prognosis

Prognosis is variable. Approximately 20% of infants with CDG-*PMM2* die within the first year of life due to severe infections, liver insufficiency, or cardiomyopathy. Some affected individuals may live into adulthood. Physical and occupational therapy can improve the quality of life.

Treatment

Treatment is symptomatic as there is no cure for CDG-*PMM2*. Treatment includes gastrostomy tubes or a specialized diet during infancy to ensure maximum calorie intake. Early intervention, physical therapy, occupational therapy, and speech therapy can also be beneficial for affected individuals. Medications may also be prescribed to help treat seizures.

Resources

Foundation Glycosylation
http://www.thefog.ca/main.html
Genetic and Rare Diseases Information Center (GARD)
https://rarediseases.org/rare-diseases/pmm2-cdg/
National Society of Genetic Counselors
https://www.nsgc.org/

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