NxGen MDx

Congenital Disorder of Glycosylation, *MPI*-Related

What Your Results Mean

Test results indicate that you are a carrier of congenital disorder of glycosylation, *MPI*-related (CDG-*MPI*). Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of congenital disorder of glycosylation, *MPI*-related 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 congenital disorder of glycosylation, *MPI*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Congenital Disorder of Glycosylation, *MPI*-Related Explained

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

Congenital disorder of glycosylation, *MPI*-related (CDG-*MPI*) is an inherited metabolic condition that affects multiple organ systems of the body. Signs and symptoms typically appear during infancy. Infants may experience extremely low blood sugar, cyclic vomiting, failure to grow, chronic diarrhea, and impaired blood clotting. Unlike other types of CDG, affected individuals that survive infancy typically do not have intellectual disability.

Prognosis

Prognosis is considered favorable with treatment, though outcomes depend on disease severity and response to medication. If left untreated, CDG-*MPI* can be fatal.

Treatment

Treatment of CDG-*MPI* consists of lifelong and daily oral supplements of mannose, a sugar. Mannose improves many of the symptoms present in this condition.

Resources

Foundation Glycosylation
http://www.thefog.ca/main.html
Genetic and Rare Diseases Information Center (GARD)
https://rarediseases.info.nih.gov/diseases/9830/mpi-cdg-cdg-ib
National Society of Genetic Counselors
https://www.nsgc.org/

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