Genetics Uncoded:

Clarity Genetics

Facts about

Congenital Disorder of Glycosylation, Type Ib

What Your Test Results Mean

Carriers typically show no symptoms of glycosylation, type Ib (CDG-Ib); however, carriers are at an increased risk of having a child with CDG-Ib. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Congenital Disorder of Glycosylation, Type Ib Explained

CDG-Ib is an inherited condition that is caused by abnormalities in mannose-6 phosphate isomerase. This enzyme is important in glycosylation, the process of adding carbohydrate chains to proteins used by the body to control protein folding, localization, activity, and stability. These carbohydrate structures cannot be formed properly without mannose-6 phosphate isomerase, leading to the development of chronic diarrhea, impaired ability of the intestines to absorb protein, and impaired blood clotting. Oral administration of mannose has been shown to be an effective treatment for this disorder.

How the Genetics Work

CDG-Ib is an autosomal recessive disorder caused by variants in the *MPI* gene. In general, individuals have two copies of the *MPI* gene. Carriers of CDG-Ib have a single variant in one copy of the *MPI* gene while individuals with CDG-Ib 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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