Genetics Uncoded:
FACTS ABOUT CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IA
AKA | Carbohydrate-Deficient Glycoprotein Syndrome, Type 1A, Phosphomannomutase 2 Deficiency, CDG-Ia

**What Your Test Results Mean**
Testing results indicate that you are a carrier of congenital disorder of glycosylation, type la (CDG-Ia). Carriers typically show no symptoms of CDG-Ia; however, carriers are at an increased risk of having a child with CDG-Ia. 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.

**Congenital Disorder of Glycosylation, Type Ia Explained**
CDG-Ia is an inherited metabolic condition that is caused by abnormalities in phosphomannomutase-2, an enzyme that is important in glycosylation of proteins. Carbohydrate chains (glycosylation) are attached to many proteins as they are produced in the cell to control protein folding, localization, activity, and stability. These carbohydrate structures cannot be formed properly without phosphomannomutase-2, leading to symptoms in multiple body systems. Symptoms of CDG-Ia usually present in the neonatal period and include global brain dysfunction, poor muscle tone, abnormal eye movement, and psychomotor retardation. Approximately 20% of infants with the disorder die within the first year of life due to severe infections, liver insufficiency, or cardiomyopathy. No treatment is currently available for this disorder.

**How the Genetics Work**
CDG-Ia is an autosomal recessive disorder caused by mutations in the PMM2 gene. In general, individuals have two copies of the PMM2 gene. Carriers of CDG-Ia have a single mutation in one copy of the PMM2 gene while individuals with CDG-Ia 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 approximately 1/3, due to preferential transmission of mutations in the PMM2 gene.

**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.