Genetics Uncoded:

Facts about Homocystinuria

What Your Test Results Mean

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

Homocystinuria Explained

Homocystinuria is an inherited metabolic disorder characterized by nearsightedness, dislocation of the lens at the front of the eye, an increased risk of abnormal blood clotting, and osteoporosis or other skeletal abnormalities. It is caused by a deficient level of the enzyme cystathionine beta-synthase. Without this enzyme, the body cannot convert homocysteine to cystathionine. As a result, homocysteine builds up in the blood. Some affected individuals experience developmental delay and intellectual disability. Symptoms of homocystinuria typically develop during the first year of life and are managed by metabolic physician and dietician.

How the Genetics Work

Homocystinuria is an autosomal recessive disorder caused by pathogenic variants in the *CBS* gene. In general, individuals have two copies of the *CBS* gene. Carriers of homocystinuria have a single variant in one copy of the *CBS* gene, while individuals with homocystinuria have variants in both copies of the *CBS* gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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