Homocystinuria Due to Cystathionine Beta-Synthase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of homocystinuria due to cystathionine beta-synthase deficiency. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of homocystinuria due to cystathionine beta-synthase deficiency 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 homocystinuria due to cystathionine beta-synthase deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Homocystinuria Due to Cystathionine Beta-Synthase Deficiency Explained

What is Homocystinuria Due to Cystathionine Beta-Synthase Deficiency?

Homocystinuria due to cystathionine beta-synthase deficiency 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. Some affected individuals experience developmental delay and intellectual disability. Symptoms of homocystinuria due to cystathionine beta-synthase deficiency vary in severity and age of onset. The condition 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.

Prognosis

Prognosis varies but is typically unfavorable. Affected individuals require careful management to minimize intellectual disability, slow the rate of lens dislocation, and reduce the incidence of seizures. The B6 nonresponsive form of homocystinuria tends to be more severe and does not improve with vitamin B6 supplementation. At age 30, affected individuals with the B6 nonresponsive form of homocystinuria have an approximate 25% chance of mortality.

Treatment

There is no cure for homocystinuria due to cystathionine beta-synthase deficiency. Treatment for patients with B6 nonresponsive homocystinuria involves dietary restriction of methionine. Individuals who are B6 responsive should follow a methionine-restricted diet and receive folate and vitamin B12 supplementation. Oral contraceptives should be avoided as they increase the risk of blood clots.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/homocystinuria

National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

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