NxGen MDx

Homocystinuria, Cobalamin E Type

What Your Results Mean

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

Homocystinuria, Cobalamin E Type Explained

What is Homocystinuria, Cobalamin E Type?

Homocystinuria, cobalamin E type is an inherited disorder in which the body is unable to process certain building blocks of proteins properly. Affected individuals will begin to show symptoms in early childhood. Symptoms include failure to grow and gain weight at the expected rate, weak muscles, megaloblastic anemia, developmental delay, and cerebral atrophy with white matter abnormalities. If left untreated, individuals may develop abnormal eye movements, abnormal gait, and seizures.

Prognosis

Prognosis is favorable with early diagnosis and treatment.

Treatment

Treatment for homocystinuria, cobalamin E type involves intramuscular cobalamin injections.

Resources

Orphanet
https://www.orpha.net

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

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

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