Methylmalonic Acidemia with Homocystinuria, Cobalamin C Type

NxGen MDx

What Your Results Mean

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

Methylmalonic Acidemia with Homocystinuria, Cobalamin C Type Explained

What is Methylmalonic Acidemia With Homocystinuria, Cobalamin C Type?

Methylmalonic acidemia with homocystinuria is an inherited disorder in which the body is unable to properly process protein building blocks (amino acids), certain fats, and cholesterol.

When the condition begins early in life, affected individuals typically have an inability to grow and gain weight at the expected rate, which is sometimes recognized before birth. These infants can also have difficulty feeding and an abnormally pale appearance (pallor). Neurological problems are also common in methylmalonic acidemia with homocystinuria, including weak muscle tone and seizures. Most infants and children with this condition have an unusually small head size, delayed development, and intellectual disability. Less common features of the condition include eye problems and a blood disorder called megaloblastic anemia. Megaloblastic anemia occurs when a person has a low number of red blood cells (anemia), and the remaining red blood cells are larger than normal (megaloblastic). The signs and symptoms of methylmalonic acidemia with homocystinuria worsen over time, and the condition can be life-threatening if not treated.

When methylmalonic acidemia with homocystinuria begins in adolescence or adulthood, the signs and symptoms usually include psychiatric changes and cognitive problems. Affected individuals can exhibit changes in their behavior and personality; they may become less social and may experience hallucinations, delirium, and psychosis. In addition, these individuals can begin to lose previously acquired mental and movement abilities, resulting in a decline in school or work performance, difficulty controlling movements, memory problems, speech difficulties, a decline in intellectual function (dementia), or an extreme lack of energy.

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Prognosis

Prognosis is poor for the early onset cases as they often experience early death. Prognosis is better with a later onset of disease.

Treatment

Treatment for methylmalonic acidemia with homocystinuria, cobalamin C type includes intramuscular injections of hydroxocobalamin, oral betaine, and folic acid. Good metabolic control and correction of hematologic problems can sometimes be achieved with this treatment, but most affected individuals continue to have signs of motor and language delay and intellectual deficit. Early diagnosis and treatment are likely most beneficial.

Resources
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia-with-homocystinuria#resources
Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/3579/methylmalonic-acidemia-with-homocystinuria
National Society of Genetic Counselors
https://www.nsgc.org/

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