Homocystinuria Due to *MTHFR* Deficiency

NxGen MDx

What Your Results Mean

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

Homocystinuria Due to *MTHFR* Deficiency Explained

What is Homocystinuria Due to *MTHFR* Deficiency?

Homocystinuria due to *MTHFR* deficiency is an inherited disorder characterized by increased levels of homocysteine in the blood and urine. Infants typically do not show symptoms at birth, but if left untreated they may develop low muscle tone, developmental delays, seizures, failure to grow and gain weight at the expected rate, blood clots, and difficulty coordinating movements (ataxia).

There are very common gene variants (C677T and A1298C) that can cause some decrease in MTHFR enzyme function. Individuals with homocystinuria due to *MTHFR* deficiency tend to have two rare variants or sometimes a rare variant in addition to a common variant. Very rarely people inherit a combination of three or four common variants from their parents (for example two C677T variants and two A1298C variants) and may also develop very high levels of homocysteine in their body.

Prognosis

Prognosis can vary depending upon the severity of the MTHFR deficiency. With early diagnosis and treatment, prognosis is typically favorable.

Treatment

Treatment involves taking betaine, folinic acid, vitamins B6 and B12, methionine, and methyltetrahydrofolate supplements as prescribed by a provider.

Resources

Orphanet
https://www.orpha.net/

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/2734/homocystinuria-due-to-mthfr-deficiency

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

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