NxGen MDx

Holocarboxylase Synthetase Deficiency

What Your Results Mean

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

Holocarboxylase Synthetase Deficiency Explained

What is Holocarboxylase Synthetase Deficiency?

Holocarboxylase synthetase deficiency is an inherited disorder in which the body is unable to use the vitamin biotin effectively. The signs and symptoms of holocarboxylase synthetase deficiency typically appear within the first few months of life, but the age of onset varies. Affected infants often have difficulty feeding, breathing problems, a skin rash, hair loss (alopecia), and a lack of energy (lethargy). If left untreated, the disorder can lead to delayed development, seizures, and coma. These medical problems may be life-threatening in some cases.

Prognosis

Prognosis is typically favorable with early diagnosis and lifelong treatment; however, some affected individuals may have lifelong learning problems. Without treatment, mortality for holocarboxylase synthetase deficiency is high.

Treatment

Treatment for holocarboxylase synthetase deficiency is free biotin supplementation as prescribed by a provider.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/holocarboxylase-synthetase-deficiency#resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/2721/holocarboxylase-synthetase-deficiency

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

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