NxGen MDx

Beta-Ketothiolase Deficiency

What Your Results Mean

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

Beta-Ketothiolase Deficiency Explained

What is Beta-Ketothiolase Deficiency?

Beta-ketothiolase deficiency is an inherited disorder in which the body cannot effectively process a protein building block (amino acid) called isoleucine. This disorder also impairs the body's ability to process ketones, which are molecules produced during the breakdown of fats.

The signs and symptoms of beta-ketothiolase deficiency typically appear between the ages of six months and 24 months. Affected children experience episodes of vomiting, dehydration, difficulty breathing, extreme tiredness, and occasionally seizures. These episodes, which are called ketoacidotic attacks, sometimes lead to coma. Ketoacidotic attacks are frequently triggered by infections, periods without food (fasting), or increased intake of protein-rich foods.

Prognosis

Prognosis depends on the severity of symptoms. In many cases, prognosis is considered favorable with early diagnosis and immediate treatment.

Treatment

L-carnitine supplementation can be considered for some individuals. Glucose and bicarbonate via IV can additionally be used during periods of a metabolic crisis. Fasting should be avoided.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/beta-ketothiolase-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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