3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency, *MCCC2*-Related

What Your Results Mean

Test results indicate that you are a carrier of 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency, *MCCC2*-related. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of 3-MCC 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 3-MCC deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency- *MCCC2*-Related Explained

What is 3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency, *MCCC2*-Related?

3-MCC deficiency is an inherited disorder in which the body is unable to process proteins containing a particular building block (amino acid) called leucine. Symptoms typically present in infancy or early childhood and consist of feeding difficulties, recurrent episodes of vomiting and diarrhea, excessive tiredness (lethargy), and weak muscle tone (hypotonia). If untreated, this disorder can lead to delayed development, seizures, and coma. The severity of these symptoms ranges from mild to life-threatening, and some people with gene variants that cause 3-MCC deficiency never experience any signs or symptoms of the condition.

Prognosis

Most individuals diagnosed with 3-MCC deficiency can live healthy lives and never need any special treatment. For individuals who do develop symptoms, prognosis is good with early and careful treatment. If left untreated, babies can develop breathing problems, seizures, liver failure, and coma.

Treatment

Treatment for 3-MCC deficiency includes avoiding fasting, a low-protein diet, and appropriate supplements (L-carnitine).

Resources

Organic Acidemia Association
https://www.oaanews.org/3-mcc.html
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/3-methylcrotonyl-coa-carboxylase-deficiency#resources
National Society of Genetic Counselors
https://www.nsgc.org/

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