NxGen MDx

Maple Syrup Urine Disease Type 1A/1B

What Your Results Mean

Test results indicate that you are a carrier of maple syrup urine disease type 1A/1B (MSUD). Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of MSUD 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 MSUD, each of your children has a 1 in 4 (25%) chance to have the condition.

Maple Syrup Urine Disease Type 1A/1B Explained

What is Maple Syrup Urine Disease Type 1A/1B?

MSUD is an inherited metabolic disorder named for the characteristic maple syrup smell in the urine of affected individuals. It is due to the lack of an enzyme responsible for processing certain amino acids (branched chain amino acids) properly. The disease is characterized by irritability, poor feeding, lethargy, seizures, and coma in infancy if untreated. Adolescents and adults with MSUD are at increased risk for ADHD, depression, and anxiety disorders. With appropriate medical management, normal growth and development are possible.

Prognosis

Prognosis is dependent upon promptness of diagnosis, management, response to diet, and episodes of metabolic crisis. Some individuals are not responsive to the recommended diet and still experience symptoms. Any neurologic symptoms and/or intellectual disabilities caused by a delay in treatment cannot be reversed. Periods of stress or infection can trigger a life-threatening metabolic crisis, which are associated with significant morbidity and mortality.

Treatment

Treatment of individuals with MSUD consists of dietary leucine restriction, branched chain amino acid-free medical foods, supplementation with isoleucine and valine, and frequent clinical and biochemical monitoring. For classic MSUD, liver transplant is also an effective therapy. Transplant eliminates dietary restrictions and provides protection from decompensations during illness.

Resources

MSUD Family Support Group
http://www.msud-support.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/maple-syrup-urine-disease
National Society of Genetic Counselors
https://www.nsgc.org/

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