Genetics Uncoded:

Facts about

Maple Syrup Urine Disease (MSUD)

What Your Test Results Mean

Carriers typically show no symptoms of maple syrup urine disease (MSUD); however, carriers are at an increased risk of having a child with MSUD. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

MSUD Explained

MSUD is an inherited metabolic disorder named for the characteristic maple syrup smell in the urine of individuals with MSUD. 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.

Individuals with MSUD do not produce enough of one of the enzymes — branched-chain alpha-ketoacid dehydrogenase — needed to metabolize branched-chain amino acids. The build-up of branched-chain amino in tissues and plasma cause signs of the disease. With appropriate medical management, normal growth and development are possible.

Treatment of individuals with MSUD consists of dietary leucine restriction, branched chain amino acid-free medical foods in addition to 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.

How the Genetics Work

The clinical features of MSUD can be explained by pathogenic variants in one of three genes — *BCKDHA*, *BCKDHB*, or *DBT*. At least 80% of MSUD is caused by variants in *BCKDHA* or *BCKDHB*. The remaining 20% of MSUD is caused by variants in the *DBT* gene. Individuals have two copies of each of the genes causing MSUD. Carriers of MSUD have a single variant in one of the MSUD genes while individuals with MSUD have variants in both copies of one of the MSUD genes, one inherited from each parent.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

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