Genetics Uncoded:

Facts about

Dihydrolipoamide Dehydrogenase Deficiency (DLD)

What Your Test Results Mean

Carriers show no symptoms of dihydrolipoamide dehydrogenase deficiency (DLD), also referred to as Maple Syrup Urine Disease (MSUD) Type III, and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

DLD Explained

DLD is an inherited metabolic disorder characterized by recurrent episodes of vomiting, abdominal pain, and hepatomegaly with or without neurologic symptoms. Onset ranges from infancy to adulthood with limited lifespan in the infantile onset form. Individuals with DLD do not generate enough of the enzyme dihydrolipoamide dehydrogenase. Deficiency of this enzyme results in a build up of amino acids and their byproducts in the body, particularly in the nervous system. Dihydrolipoamide dehydrogenase is also required to converting pyruvate into ATP, a primary source of energy. Without dihydrolipoamide dehydrogenase, the body is not able to produce enough energy. Unlike classic MSUD, lifelong restriction of amino acids is not always effective for treating DLD. Treatment of individuals with DLD is supportive.

How the Genetics Work

DLD is an autosomal recessive disorder caused by pathogenic variants in the *DLD* gene. In general, individuals have two copies of the *DLD* gene. Carriers of DLD have a single variant in one copy of the *DLD* gene while individuals with DLD have variants in both copies of their genes. Risk for two carriers to have a child with the disorder is 25%.

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.

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