Genetics Uncoded:

NxGen MDx

Facts about
Glutaric Acidemia Type Ia

What Your Test Results Mean

Carriers show no symptoms of glutaric acidemia type Ia 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.

Glutaric Acidemia Type Ia Explained

Glutaric acidemia type Ia is an inherited metabolic disorder caused by a missing enzyme, glutaryl-CoA dehydrogenase. Without this enzyme, the body cannot break down the amino acids lysine and tryptophan. Symptoms may include metabolic crisis (severe vomiting and lethargy that can lead to seizures and other complications) in times of illness, fever, or fasting. To avoid a metabolic crisis, individuals with the disorder should avoid fasting and may be placed on a special diet. Diet and medications for individuals with glutaric acidemia type Ia are typically managed by a metabolic physician and dietician. With proper management, most individuals with glutaric acidemia type Ia will live long and healthy lives.

How the Genetics Work

Glutaric acidemia type Ia is an autosomal recessive disorder caused by pathogenic variants in the *GCDH* gene. In general, individuals have two copies of the *GCDH* gene. Carriers of glutaric acidemia type Ia have a single variant in one copy of the *GCDH* gene while individuals with glutaric acidemia type Ia have variants in both copies of their genes, one inherited from each parent. 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.

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

Revised 12/15/16