Genetics Uncoded:

NxGen MDx

Facts about Galactosemia

What Your Test Results Mean

Carriers do not show symptoms of galactosemia 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.

Galactosemia Explained

Galactosemia is an inherited metabolic disorder that prevents the body from processing galactose, a simple sugar. If infants are not promptly treated with a low galactose diet, symptoms and complications can occur days after birth. Feeding difficulties, lethargy, failure to thrive and jaundice, as well as sepsis (a severe bacterial infection) and shock, may be seen in untreated individuals. Affected females may develop ovarian failure. Children with this disorder are at in increased risk for cataracts, delayed development, speech difficulties, and intellectual disabilities. Those with this condition must alter their diet to avoid all milk and milk-containing products to avoid galactose. Antibiotics can be effective in preventing sepsis.

How the Genetics Work

Galactosemia is an autosomal recessive disorder caused by pathogenic variants in the *GALT* gene. In general, individuals have two copies of the *GALT* gene. Carriers of galactosemia have a variant in one copy of the *GALT* gene while individuals with galactosemia have variants in both copies of the *GALT* gene, 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.

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