NxGen MDx

Galactosemia, *GALT*-Related

What Your Results Mean

Test results indicate that you are a carrier of galactosemia, *GALT*-related. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of galactosemia, *GALT*-related 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 galactosemia, *GALT*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Galactosemia, *GALT*-Related Explained

What is Galactosemia?

Galactosemia, *GALT*-related is an inherited metabolic disorder that prevents the body from processing galactose, a simple sugar, into energy. 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, jaundice, sepsis (a severe bacterial infection), and shock may be seen in untreated individuals. Even with treatment, children with galactosemia are more likely to develop cataracts, delayed development, speech difficulties, and intellectual disabilities. Affected females may develop ovarian failure.

Prognosis

Prognosis is variable. Individuals with the condition must alter their diet to avoid all milk and milk-containing products to avoid galactose. Maintaining a galactose-free diet can prevent or improve liver disease, lethargy, vomiting, diarrhea, cataracts, and the risk of sepsis. However, developmental delay, intellectual disability, speech problems, premature ovarian failure, and movement difficulties may be present even in individuals who maintain a galactose-restricted diet.

Treatment

Treatment involves a lifelong galactose-free diet (avoiding all milk products) beginning as early in life as possible. Vitamin supplementation with calcium, vitamin K, and vitamin D is typically recommended for bone health. Speech therapy may help address speech delay and impaired speech.

Resources

Galactosemia Foundation
http://www.galactosemia.org/about/
National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/galactosemia/
National Society of Genetic Counselors
https://www.nsgc.org/

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