Galactosemia, *GALE*-Related

NxGen MDx

What Your Results Mean

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

Galactosemia, *GALE*-Related Explained

What is Galactosemia?

Galactosemia, *GALE*-related is an inherited metabolic disorder that prevents the body from processing galactose, a simple sugar, into energy. In generalized galactosemia, *GALE*-related, 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, an enlarged liver, aminoaciduria, intellectual disabilities, and cataracts may be seen in untreated individuals. In the peripheral and intermediate forms, infants may remain clinically well even on a regular milk diet and are usually only identified by biochemical testing, often in newborn screening programs.

Prognosis

Prognosis is variable. Individuals with the condition must alter their diet to restrict most milk and milk-containing products to avoid galactose. Maintaining a galactose-free diet can prevent or improve liver disease, lethargy, vomiting, diarrhea, and cataracts. However, developmental delay, intellectual disability, and/or speech problems may be present even in individuals who maintain a galactose-restricted diet. Prognosis is typically good for individuals with the peripheral or intermediate forms, as they are often asymptomatic and do not require treatment.

Treatment

Treatment involves a lifelong galactose-restricted diet beginning as early in life as possible. Persons with galactosemia, *GALE*-related may require trace environmental sources of galactose. However, the galactose intake needed for optimum outcome remains unknown. Vitamin supplementation with calcium, vitamin K, and vitamin D is typically recommended for bone health. Individuals with the peripheral or intermediate forms may not require any treatment.

Resources

Galactosemia Foundation
http://www.galactosemia.org/about/

GeneReviews
https://www.ncbi.nlm.nih.gov/books/NBK51671/#gale-def.Management

National Society of Genetic Counselors
https://www.nsgc.org/

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Revised: 7/2020