

Galactokinase Deficiency

What Your Results Mean

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

Galactokinase Deficiency Explained

What is Galactokinase Deficiency?

Galactokinase deficiency is an inherited metabolic disorder that prevents the body from processing galactose, a simple sugar, into energy. It is typically considered a mild type of galactosemia as affected children who are not treated with dietary therapy develop cataracts in the eyes, but otherwise they typically do not have long-term health problems. Occasionally, a child with galactokinase deficiency will have pseudotumor cerebri, a condition which mimics the symptoms of a large brain tumor when no brain tumor is present.



Prognosis

Prognosis is good with treatment. Individuals who receive an early diagnosis and treatment can reduce the complications of vision problems.

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.



Resources

Genetic and Rare Diseases Information Center

<https://rarediseases.info.nih.gov/diseases/2422/galactokinase-deficiency>

GeneReviews

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

National Society of Genetic Counselors

<https://www.nsgc.org/>