NxGen MDx

Familial Hyperinsulinism, *ABCC8*-Related

What Your Results Mean

Test results indicate that you are a carrier of familial hyperinsulinism, *ABCC8*-related. Some carriers show no symptoms of this condition, while others have symptoms ranging from congenital hyperinsulinism to type 2 diabetes mellitus in adulthood. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

We recommend that you share this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

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 are carriers for familial hyperinsulinism, *ABCC8*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Familial Hyperinsulinism, *ABCC8*-Related Explained

What is Familial Hyperinsulinism, *ABCC8*-Related?

Familial hyperinsulinism is an inherited disorder characterized by hypoglycemia (low blood sugar). Individuals with familial hyperinsulinism release insulin into the bloodstream even in the absence of glucose. In infants and young children, these episodes of low blood sugar are characterized by a lack of energy, irritability, and difficulty feeding. Repeated episodes of low blood sugar increase the risk for seizures, intellectual disability, breathing difficulties, and coma.

Carriers for familial hyperinsulinism may be at increased risk for persistent hyperinsulinemic hypoglycemia in childhood. Additionally, it may be an important cause of dominantly inherited early-onset diabetes mellitus in adults.

Prognosis

Prognosis with treatment is variable. In severe cases, surgical removal of parts of the pancreas is required to control the release of insulin. However, this surgery increases the risk of developing diabetes and neurological deficits from episodes of low blood sugar. In mild cases, patients typically respond well to medication. Even with treatment, some individuals may have some degree of brain damage or learning disabilities. Overall, early diagnosis and immediate treatment are associated with the best prognosis.

Treatment

There are two options for treatment of familial hyperinsulinism, medical therapy and surgical intervention. About 50% of children respond to medical therapy, while the other half require surgery for a partial or near total pancreatectomy.

Resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/3947/familial-hyperinsulinism

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/congenital-hyperinsulinism

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

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