Glycine Encephalopathy, *GLDC*-Related

NxGen MDx

What Your Results Mean

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

Glycine Encephalopathy, *GLDC*-Related Explained

What is Glycine Encephalopathy, *GLDC*-Related?

Glycine encephalopathy is caused by the shortage of an enzyme that normally breaks down glycine in the body. A lack of this enzyme allows excess glycine to build up in tissues and organs, particularly the brain, leading to serious medical problems. The most common form of glycine encephalopathy, called the classical type, appears shortly after birth. Affected infants experience a progressive lack of energy, feeding difficulties, weak muscle tone, abnormal jerking movements, and life-threatening problems with breathing. Most children who survive these early signs and symptoms develop profound intellectual disability and seizures that are difficult to treat. For unknown reasons, affected males are more likely to survive and have less severe developmental problems than affected females.

Several other types of glycine encephalopathy with variable signs and symptoms have been identified. The most common of these atypical types is called the infantile form. Children with this condition develop normally until they are about six months old, when they experience delayed development and may begin having seizures. As they get older, many develop intellectual disability, abnormal movements, and behavioral problems. Other atypical types of glycine encephalopathy appear later in childhood or adulthood and cause a variety of medical problems that primarily affect the nervous system.

Prognosis

Prognosis is typically poor for the classic type of glycine encephalopathy, *GLDC*-related. About half of the infants with the classic form die within a few weeks of life, and survivors may have motor delay, profound intellectual disability, seizures, and stiffness. Prognosis for the later onset types varies depending on the age of onset of symptoms and the degree of enzyme deficiency.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022

Treatment

Treatment for glycine encephalopathy, *GLDC*-related is supportive and symptomatic. Affected individuals may require antiepileptics for seizure control, placement of a gastrostomy tube for swallowing disorders, and treatment for gastroesophageal reflux. Sodium benzoate is used to reduce plasma glycine levels.

Resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/7219/glycine-encephalopathy

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/glycine-encephalopathy#genes

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

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022