NxGen MDx

Lysosomal Acid Lipase Deficiency

What Your Results Mean

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

Lysosomal Acid Lipase Deficiency Explained

What is Lysosomal Acid Lipase Deficiency?

Lysosomal acid lipase deficiency is an inherited condition characterized by problems with the breakdown and use of fats and cholesterol in the body. In affected individuals, harmful amounts of fats accumulate in cells and tissues throughout the body, which typically causes liver disease. There are two forms of the condition: the most severe and rarest form begins in infancy, while the less severe form can begin from childhood to late adulthood.

In the severe, early-onset form, lipids accumulate throughout the body within the first weeks of life, particularly in the liver. This accumulation of lipids leads to several health problems, including an enlarged liver and spleen, poor weight gain, jaundice, vomiting, diarrhea, fatty stool, and poor absorption of nutrients from food (malabsorption). In addition, affected infants often have calcium deposits in their adrenal glands, low amounts of iron in the blood (anemia), and developmental delay. Scar tissue quickly builds up in the liver, leading to liver disease (cirrhosis).

In the later-onset form of lysosomal acid lipase deficiency, signs and symptoms vary and usually begin in mid-childhood, although they can appear anytime up to late adulthood. Nearly all affected individuals develop an enlarged liver; an enlarged spleen may also occur. About two-thirds of individuals have liver fibrosis, eventually leading to cirrhosis. Approximately one-third of individuals with the later-onset form have malabsorption, diarrhea, vomiting, and steatorrhea.

Some people with this later-onset form develop an accumulation of fatty deposits on the artery walls. The deposits narrow the arteries, increasing the chance of heart attack or stroke.

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

Prognosis

Prognosis for the severe, early-onset form is poor, as infants with this form of lysosomal acid lipase deficiency develop multi-organ failure and severe malnutrition and generally do not survive past one year. Prognosis varies for the later-onset form as the expected lifespan depends on the severity of the associated health problems.

Treatment

Treatment includes supportive measures that aim to control cholesterol levels and liver damage through medications and diet. Enzyme replacement therapy (ERT) with the enzyme sebelipase alfa is available; however, the long-term clinical efficacy is yet to be determined.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/lysosomal-acid-lipase-deficiency#inheritance

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