Lipoid Congenital Adrenal Hyperplasia, *STAR*-Related

NxGen MDx

What Your Results Mean

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

Lipoid Congenital Adrenal Hyperplasia, *STAR*-Related Explained

What is Lipoid Congenital Adrenal Hyperplasia, *STAR*-Related?

Congenital adrenal hyperplasia (CAH) is a group of rare inherited disorders characterized by a deficiency of one of the enzymes needed to make specific hormones. Lipoid congenital adrenal hyperplasia, *STAR*-related (LCAH) is the most severe form of congenital adrenal hyperplasia. Individuals with this type of CAH lack the ability to make almost all of the adrenal and gonadal steroid hormones.

In the classic type of LCAH, individuals usually experience salt-wasting crises in early infancy, where the body cannot retain salt leading to dehydration and other complications that can be life-threatening. They may also be lethargic, have poor feeding, and almost all affected individuals have female genitalia. Some neurologic abnormalities have also been described.

In the non-classic type of LCAH, which has been described as a form of non-autoimmune Addison disease, symptoms tend to present in infancy or early childhood. Individuals may experience salt-wasting crises, darkening of the skin, vomiting, and low-blood sugar.

Prognosis

Without treatment, prognosis for LCAH is typically fatal in early infancy. However, with early and consistent adherence to treatment, affected individuals can live into adulthood.

Treatment

Treatment for LCAH consists of hormone replacement therapy. A multidisciplinary team of specialists is often required including an endocrinologist to monitor the medication dosage, medication side effects, growth, and development (both general and sexual) of individuals who continue to receive treatment. Surgery may be considered to correct the appearance of ambiguous genitalia.

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

Resources

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/congenital-adrenal-hyperplasia/

CARES Foundation
https://caresfoundation.org/

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