3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (Congenital Adrenal Hyperplasia)

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of 3-beta-hydroxysteroid dehydrogenase type II deficiency (CAH). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of CAH 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 are carriers for CAH, each of your children has a 1 in 4 (25%) chance to have the condition.

3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (Congenital Adrenal Hyperplasia) Explained

What is 3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (CAH)?

CAH is an inherited condition that affects hormone-producing glands including the gonads (ovaries and testes) and the adrenal glands. The gonads direct sexual development before birth and during puberty. The adrenal glands, which are located on top of the kidneys, regulate the production of certain hormones and control salt levels in the body. There are three types of CAH: the salt-wasting, non-salt-wasting, and non-classic types.

In the salt-wasting type, hormone production is extremely low. Individuals with this type lose large amounts of sodium in their urine, which can be life-threatening. Individuals affected with the salt-wasting type are usually diagnosed soon after birth due to complications related to a lack of salt reabsorption, including dehydration, poor feeding, and vomiting. Individuals with the non-salt-wasting type of CAH produce enough hormone to allow sodium reabsorption in the kidneys. Individuals with the non-classic type have the mildest symptoms and do not experience salt wasting.

In males with any type of CAH, problems with male sex hormones lead to abnormalities of the external genitalia. These abnormalities range from having the opening of the urethra on the underside of the penis (hypospadias) to having external genitalia that do not look clearly male or female (ambiguous genitalia). Affected males are also frequently infertile.

Females with CAH may have slight abnormalities of the external genitalia at birth. Females affected with the non-salt-wasting or non-classic types are typically not diagnosed until mid-childhood or puberty, when they may experience irregular menstruation, premature pubic hair growth, and excessive body hair growth. Females with CAH also have impaired fertility.

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Revised: 4/2022

Prognosis

The long-term prognosis for people with CAH is usually good-to-excellent with adequate hormone replacement therapy monitoring.

Treatment

Treatment may include hormone replacement therapy which is typically monitored by an endocrinologist. Individuals with ambiguous genitalia may require surgery to correct the function and appearance of the external genitalia.

Resources

CARES Foundation
http://www.caresfoundation.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/congenital-adrenal-hyperplasia-due-to-11-beta-hydroxylase-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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