NxGen MDx

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

What Your Results Mean

Test results indicate that you are a carrier of 11-beta-hydroxylase-deficient congenital adrenal hyperplasia (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.

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia Explained

What is 11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CAH)?

CAH is an inherited condition that affects the adrenal glands. The role of the adrenal glands is to produce hormones that regulate many essential body functions. In CAH, the adrenal glands produce excess androgens, which are male sex hormones. There are two major types of CAH: classic CAH and non-classic CAH.

Classic CAH is characterized by ambiguous external genitalia in females (not clearly male or female), though the internal reproductive organs typically develop normally. Males and females with the classic form of this condition have early development of their secondary sexual characteristics such as growth of facial and pubic hair, deepening of the voice, appearance of acne, and onset of a growth spurt (though their adult height is typically shorter than average). Additionally, two-thirds of affected individuals will develop hypertension within the first few years of life.

The non-classic type is less severe than classic CAH. Females typically have normal genitalia, though they may develop excessive body hair growth (hirsutism) and irregular menstruation as they get older. Males with the non-classic type are often asymptomatic aside from short stature.

Prognosis

Early diagnosis and proper management allow for a normal life expectancy. Issues with hypertension, growth, development, infertility, and ambiguous genitalia will need to be monitored by a physician.

Treatment

Treatment may include hormone replacement therapy which is typically monitored by an endocrinologist. Females with ambiguous genitalia may require surgery to correct the function and appearance of the external genitalia. Those individuals with hypertension will require antihypertensive therapy.

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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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