21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of 21-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.

21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CAH) Explained

What is 21-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. With 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 can be divided into two types: the salt-wasting type and the simple virilizing type. In the classic salt-wasting type, individuals are unable to retain enough salt, leading to dehydration, vomiting, diarrhea, failure to thrive, heart rhythm abnormalities, and shock. Additionally, affected males may present at birth with enlarged genitalia, and affected females may have ambiguous genitalia (not appearing as male or female). Other symptoms include an early growth spurt but overall shorter than average adult height, infertility, abnormal menstruation cycles for females, excess facial hair for females, and early facial hair growth for males.

In the classic simple virilizing type, affected individuals have similar symptoms as the salt-wasting type, except these individuals typically do not experience severe and life-threatening sodium deficiency symptoms as newborns.

The non-classic type is less severe than classic CAH. Individuals with non-classic CAH may not present with symptoms until childhood, adolescence, or adulthood. Some individuals may even remain asymptomatic throughout their life. Those affected may experience an early growth spurt with shorter than average adult height and infertility. Additionally, girls may experience symptoms of masculinization and abnormal menstruation.

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Prognosis

Early diagnosis and proper management allow for a normal life expectancy. If left untreated or not properly managed, salt-wasting crises can become life-threatening. Issues with growth and 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.

Resources

CARES Foundation
http://www.caresfoundation.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/21-hydroxylase-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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