NxGen MDx

17-Alpha-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

What Your Results Mean

Test results indicate that you are a carrier of 17-alpha-hydroxylase-deficient congenital adrenal hyperplasia (CAH). 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.

17-Alpha-Hydroxylase-Deficient Congenital Adrenal Hyperplasia Explained

What is CAH?

CAH is a condition that affects the function of certain hormone-producing glands called the gonads and the adrenal glands. The gonads direct sexual development before birth and during puberty and are important for reproduction. The adrenal glands, which are located on top of the kidneys, regulate the production of certain hormones, including those that control salt levels in the body. Hormone imbalances lead to the characteristic signs and symptoms of CAH, which include high blood pressure, low levels of potassium in the blood, and abnormal sexual development. Severity and symptoms differ based on the biological sex of the affected individual and if the individual has complete or partial CAH.

Females with CAH are born with normal external female genitalia; however, the internal reproductive organs, including the uterus and ovaries, may be underdeveloped. Women with complete CAH do not develop secondary sex characteristics, such as breasts and pubic hair, and do not menstruate. Women with partial CAH may develop some secondary sex characteristics; menstruation is typically irregular or absent. Either form of the disorder results in an inability to conceive a baby.

Males with CAH have problems with sexual development that lead to abnormalities of the external genitalia. The most severely affected are born with characteristically female external genitalia and are generally raised as females. However, because they do not have female internal reproductive organs, these individuals have amenorrhea and do not develop female secondary sex characteristics. These individuals have testes, but they are abnormally located in the abdomen (undescended). Sometimes, complete CAH leads to external genitalia that do not look clearly male or clearly female (ambiguous genitalia). Males with partial CAH usually have abnormal male genitalia, such as a small penis, the opening of the urethra on the underside of the penis, or a scrotum divided into two lobes. Males with either complete or partial CAH are also infertile.

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Prognosis

Early diagnosis and proper management allow for a normal life expectancy. Issues with hypertension, 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. Males with ambiguous genitalia may require surgery to correct the function and appearance of the external genitalia. Those individuals with hypertension will require antihypertensive therapy.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/17-alpha-hydroxylase-17-20-lyase-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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