

Combined Pituitary Hormone Deficiency

What Your Results Mean

Test results indicate that you are a carrier of combined pituitary hormone deficiency. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of combined pituitary hormone deficiency 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 combined pituitary hormone deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Combined Pituitary Hormone Deficiency Explained

What is Combined Pituitary Hormone Deficiency?

Combined pituitary hormone deficiency is an inherited disorder that causes a deficiency or shortage of many hormones that are produced by the pituitary gland. A deficiency of these hormones can affect the development of many parts of the body. Affected individuals typically first show signs of short stature or a failure to grow at an expected rate in childhood. Additional signs and symptoms can include hypothyroidism, delayed or absent puberty, infertility, or an impaired immune system. Very rarely, affected individuals may have an intellectual disability.



Prognosis

Prognosis for individuals with combined pituitary hormone deficiency is considered good. Affected individuals typically live a normal lifespan.

Treatment

Treatment for combined pituitary hormone deficiency consists of hormone replacement therapy.



Resources

Human Growth Foundation

<http://hgfound.org/>

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/combined-pituitary-hormone-deficiency#inheritance>

National Society of Genetic Counselors

<https://www.nsgc.org/>