



Genetics Uncoded:

Facts about

Combined Pituitary Hormone Deficiency



What Your Test Results Mean

Carriers show no symptoms of combined pituitary hormone deficiency and are not at risk to develop combined pituitary hormone deficiency. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

● Combined Pituitary Hormone Deficiency Explained

Combined pituitary hormone deficiency is an inherited disorder caused by lack of pituitary hormones. Variants in the *PROP1* gene prevent the production of several hormones, such as growth hormone, luteinizing hormone, thyroid-stimulating hormone, and adrenocorticotrophic hormone. Failure to grow at the expected rate and short stature are apparent in early childhood. Affected individuals may also have hypothyroidism, which can cause weight gain and fatigue. Delayed or absent puberty and infertility may also be symptoms. Treatment includes injections of biosynthetic growth hormone, though this may induce the development of secondary sexual characteristics.

● How the Genetics Work

Combined pituitary hormone deficiency is an autosomal recessive disorder caused by variants in the *PROP1* gene. Most cases of combined pituitary hormone deficiency are sporadic, meaning variants in the *PROP1* gene have spontaneously appeared in an individual with no familial history. When the disease is familial, it can be autosomal recessive or autosomal dominant. In general, individuals have two copies of the *PROP1* gene. If the pattern is autosomal recessive, carriers of combined pituitary hormone deficiency have a single variant in one copy of the *PROP1* gene, while individuals with combined pituitary hormone deficiency have variants in both copies of the *PROP1* gene, one inherited from each parent. If the pattern is autosomal dominant, only one variant in one copy of the *PROP1* gene is necessary to be an affected individual. Risk for two carriers to have a child with the

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.