

Identify genetic causes of CPHD

Combined Pituitary Hormone Deficiency Evaluation

What is Combined Pituitary Hormone Deficiency (CPHD)?

- CPHD presents in childhood with severe short stature and deficiency in anterior pituitary hormones.
 - Growth failure usually becomes apparent between ages 2 and 8.
 - GH deficiency is recognized first with deficiencies in a second anterior pituitary hormone, usually TSH or prolactin, appearing later.
- CPHD can be familial or the result of pituitary trauma or tumors.
 - Genetic causes of CPHD include mutations in *PROP1* or *POU1F1* (also known as *PIT1*), transcription factors involved in anterior pituitary development.¹⁻³

PROP1 and *POU1F1* as Causes of CPHD⁴⁻⁵

	<i>PROP1</i>	<i>POU1F1</i>
Pituitary Hormone Deficiencies	GH, TSH, prolactin, LH, FSH, occasionally ACTH	GH, TSH, prolactin
Anterior Pituitary MRI Findings	Most patients show hypoplasia. However, anterior pituitary may be normal, and there are some reports of transient hyperplasia.	Normal or hypoplasia
Effect on Puberty	Most patients do not undergo spontaneous puberty.	Not affected
Inheritance	Autosomal Recessive	Autosomal Dominant or Autosomal Recessive
% of CPHD	30-50%	30%



Why genetic testing?

Genetic testing for mutations in *PROP1* or *POU1F1* associated with CPHD may

- Help to diagnose familial CPHD in over 50% of cases and clarify the mode of inheritance.
- Facilitate better treatment by alerting physicians and families to future expected hormone deficiencies. Starting GH therapy earlier in patients may result in increased final height.
- Help to discriminate between genetic and tumor-related CPHD.
- Allow carrier detection and early diagnosis of CPHD in affected descendants.

Indications for testing:

- Combined deficiencies in growth hormone and TSH or prolactin
- Family history of CPHD

For complete ordering information, please see the reverse side.

References: 1. Deladoey J, et al., (1999) *J Clin Endocrinol Metab* 84:1645-50. 2. Turton JPG, et al., (2005) *Clinical Endocrinology* 63:10-18. 3. Turton JPG, et al., (2005) *J Clin Endocrinol Metab* 90:4762-70. 4. Parks JS, et al., (1999) *J Clin Endocrinol Metab* 84:4362-70. 5. Reynaud R, et al., (2004) *Growth Hormone & IGF Research* 14:442-8.

For a brief review on Combined Pituitary Hormone Deficiency, please visit www.athenadiagnostics.com

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Combined Pituitary Hormone Deficiency Evaluation

- Typical Presentation:** Severe short stature and failure to thrive in young children
- Indications for Testing:**
- Combined deficiencies in growth hormone and TSH or prolactin
 - Family history of CPHD

TEST DETAILS

- Test Code:** 865
- Test Includes:** *PROP1* (CPHD) DNA Sequencing Test, #863
POU1F1 (CPHD) DNA Sequencing Test, #864
- Test Turnaround:** 21-28 days

TECHNICAL INFORMATION

- Methodology:** Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region of genes

SHIPPING CONSIDERATIONS

- Specimen Type:** Whole blood
- Volume:** 10 mL (pediatric minimum: 2 mL)
- Collection Tube:** Yellow or lavender top
- Stability:** Hemolysis may compromise DNA recovery and integrity after 48 hrs
- Storage Conditions:** For short periods (until shipped) at 4°C
- Shipping Conditions:** Overnight at room temperature (specimen arrival must be less than 24 hrs after collection); ship Monday through Thursday only

To order the Combined Pituitary Hormone Deficiency Evaluation (Test #865), call Athena Diagnostics' Customer Service Representatives at:

800-394-4493 x2



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