MAYO CLINIC LABORATORIES

Test Definition: OHPG

17-Hydroxyprogesterone, Serum

Overview

Useful For

The analysis of 17-hydroxyprogesterone (17-OHPG) is 1 of the 3 analytes along with cortisol and androstenedione, that constitutes the best screening test for congenital adrenal hyperplasia (CAH), caused by either 11- or 21-hydroxylase deficiency.

Analysis for 17-OHPG is also useful as part of a battery of tests to evaluate females with hirsutism or infertility; both can result from adult-onset CAH

Testing Algorithm

See Steroid Pathways in Special Instructions.

Special Instructions

- Steroid Pathways

Method Name

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Serum Red

Necessary Information

Patient's age and sex are required.

Specimen Required

Container/Tube: Red top **(serum gel/SST are not acceptable)**

Specimen Volume: 0.6 mL

Forms

If not ordering electronically, complete, print, and send a General Request (T239) with the specimen.

Specimen Minimum Volume

0.25 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	Reject
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum Red	Refrigerated (preferred)	28 days	
	Frozen	28 days	
	Ambient	7 days	

Clinical & Interpretive

Clinical Information

Congenital adrenal hyperplasia (CAH) is caused by inherited defects in steroid biosynthesis. The resulting hormone imbalances with reduced glucocorticoids and mineralocorticoids and elevated 17-hydroxyprogesterone (OHPG) and androgens can lead to life-threatening, salt-wasting crisis in the newborn period and incorrect gender assignment of virtualized females. Adult-onset CAH may result in hirsutism or infertility in females.

The adrenal glands, ovaries, testes, and placenta produce OHPG. It is hydroxylated at the 11 and 21 position to produce cortisol. Deficiency of either 11- or 21-hydroxylase results in decreased cortisol synthesis, and feedback inhibition of adrenocorticotropic hormone (ACTH) secretion is lost. Consequent increased pituitary release of ACTH increases production of OHPG. But, if 17-alpha-hydroxylase (which allows formation of OHPG from progesterone) or 3-beta-hydroxysteroid dehydrogenase type 2 (which allows formation of 17-hydroxyprogesterone formation from 17-hydroxypregnenolone) are deficient, OHPG levels are low with possible increase in progesterone or pregnenolone respectively.

OHPG is bound to both corticosteroid binding globulin and albumin and total OHPG is measured in this assay. OHPG is converted to pregnanetriol, which is conjugated and excreted in the urine. In all instances, more specific tests are available to diagnose disorders or steroid metabolism than pregnanetriol measurement.

Most (90%) cases of CAH are due to variants in the steroid 21-hydroxylase gene (*CYP21A2*). CAH due to 21-hydroxylase deficiency is diagnosed by confirming elevations of OHPG and androstenedione (ANST / Androstenedione, Serum) with decreased cortisol (CINP / Cortisol, Mass Spectrometry, Serum). By contrast, in 2 less common forms of CAH, due to 17-hydroxylase or 11-hydroxylase deficiency, OHPG and androstenedione levels are not significantly elevated and measurement of progesterone (PGSN / Progesterone, Serum) and deoxycorticosterone (FDOC / Deoxycorticosterone [DOC], Serum), respectively, are necessary for diagnosis.

CAH21 / Congenital Adrenal Hyperplasia (CAH) Profile for 21-Hydroxylase Deficiency allows the simultaneous determination of OHPG, androstenedione, and cortisol.

See Steroid Pathways in Special Instructions.

Reference Values

Children:
Preterm infants
Preterm infants may exceed 630 ng/dL, however, it is uncommon to see levels reach 1,000 ng/dL.
Term infants
0-28 days: <630 ng/dL
Levels fall from newborn (<630 ng/dL) to prepubertal gradually within 6 months.
Prepubertal males: <110 ng/dL
Prepubertal females: <100 ng/dL

Adults:
Males: <220 ng/dL
Females
Follicular: <80 ng/dL
Luteal: <285 ng/dL
Postmenopausal: <51 ng/dL

Note: For pregnancy reference ranges, see: Soldin OP, Guo T, Weiderpass E, et al: Steroid hormone levels in pregnancy and 1 year postpartum using isotope dilution tandem mass spectrometry. Fertil Steril. 2005 Sept;84(3):701-710

Interpretation

Diagnosis and differential diagnosis of congenital adrenal hyperplasia (CAH) always requires the measurement of several steroids. Patients with CAH due to steroid 21-hydroxylase gene (*CYP21A2*) variants usually have very high levels of androstenedione, often 5- to 10-fold elevations. 17-hydroxyprogesterone (OHPG) levels are usually even higher, while cortisol levels are low or undetectable. All 3 analytes should be tested.

In the much less common *CYP11A1* variant, androstenedione levels are elevated to a similar extent as in *CYP21A2* variant, and cortisol is also low, but OHPG is only mildly, if at all, elevated.

In the also very rare 17-alpha-hydroxylase deficiency, androstenedione, all other androgen-precursors (17-alpha-hydroxypregnenolone, OHPG, dehydroepiandrosterone sulfate), androgens (testosterone, estrone, estradiol), and cortisol are low, while production of mineral corticoid and its precursors, in particular progesterone, 11-deoxycorticosterone, and 18-hydroxycorticosterone, are increased.

The goal of CAH treatment is normalization of cortisol levels and ideally also of sex-steroid levels. Traditionally, OHPG and urinary pregnanetriol or total ketosteroid excretion are measured to guide treatment, but these tests correlate only modestly with androgen levels. Therefore, androstenedione and testosterone should also be measured and used to guide treatment modifications. Normal prepubertal levels may be difficult to achieve, but if testosterone levels are within the reference range, androstenedione levels of up to 100 ng/dL are usually regarded as acceptable.

See Steroid Pathways in Special Instructions.

Cautions

At birth the hypothalamic-pituitary-adrenal axis and the hypothalamic-pituitary-gonadal axis are activated, and adrenal and sex steroid levels are high. In preterm infants the elevations can be even more pronounced due to illness and stress. As a result, preterm infants may occasionally have 17-hydroxyprogesterone levels of up to 1000 ng/dL. Term infants

(0-28 days) will have levels less than 630 ng/dL. These then fall over the following 1 to 6 months to prepubertal levels of less than 110 ng/dL (males) and less than 100 ng/dL (females).

Clinical Reference

1. Therrell BL: Newborn screening for congenital adrenal hyperplasia. Endocrinol Metab Clin North Am. 2001;30(1):15-30
2. Bachega TA, Billerbeck AE, Marcondes JA, et al: Influence of different genotypes on 17-hydroxyprogesterone levels in patients with non-classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Clin Endocrinol. 2000;52(5):601-607
3. Von Schnaken K, Bidlingmaier F, Knorr D: 17-hydroxyprogesterone, androstenedione, and testosterone in normal children and in prepubertal patients with congenital adrenal hyperplasia. Eur J Pediatr. 1980;133(3):259-267
4. Sciarra F, Tosti-Croce C, Toscano V: Androgen-secreting adrenal tumors. Minerva Ednocrinol. 1995;20(1):63-68
5. Collett-Solberg PF: Congenital adrenal hyperplasia: from genetics and biochemistry to clinical practice, part I. Clin Pediatr. 2001;40(1):1-16
6. Soldin OP, Guo T, Weiderpass E, et al: Steroid hormone levels in pregnancy and 1 year postpartum using isotope dilution tandem mass spectrometry. Fertil Steril. 2005 Sept;84(3):701-710
7. Speiser PW, Azziz R, Baskin LS, et al: Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: An Endocrine Society Clinical Practice Guideline. J Clin Endocrinol Metab. 2010;95(9):4133-4160 Available at: jcem.endojournals.org

Performance

Method Description

17-Hydroxyprogesterone and internal standard are extracted from serum. The extract is quantified using high-performance liquid chromatography-tandem mass spectrometry (LC-MS/MS).(Wudy SA, Hartmann M, Svoboda M: Determination of 17-hydroxyprogesterone in plasma by stable isotope dilution/benchtop liquid chromatography-tandem mass spectrometry. Horm Res. 2000;53[2]:68-71)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

2 to 5 days

Specimen Retention Time

14 days

Performing Laboratory Location

Rochester

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

83498

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
OHPG	17-Hydroxyprogesterone, S	1668-3

Result ID	Test Result Name	Result LOINC® Value
9231	17-Hydroxyprogesterone, S	1668-3