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

Specimen Required
Container/Tube: Red top

Specimen Volume: 0.6 mL

Additional Information: Indicate patient's age and sex.

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

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<th>Condition</th>
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<tr>
<td>Gross lipemia</td>
<td>Reject</td>
</tr>
<tr>
<td>Gross icterus</td>
<td>OK</td>
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<td>Other</td>
<td>Serum gel tube</td>
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Specimen Stability Information
Clinical and 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 mutations 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, Serum, LC-MS/MS). 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


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) mutations 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 mutation, androstenedione levels are elevated to a similar extent as in CYP21A2 mutation, 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-hydroxyprogrenolone, 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-hydroxy cortisol, 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 1,000 ng/dL. Term infants (0-28 days) will have levels <630 ng/dL. These then fall over the following 1 to 6 months to prepubertal levels of <110 ng/dL (males) and <100 ng/dL (females).

Clinical Reference


### Performance

#### Method Description


#### PDF Report

No

#### Day(s) and Time(s) Test Performed

Monday through Friday; 4 p.m.

#### Analytic Time

2 days

#### Maximum Laboratory Time

5 days

#### Specimen Retention Time

14 days

#### Performing Laboratory Location

Rochester

#### Fees and 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 Regional Manager. 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 U.S. Food and Drug Administration.

CPT Code Information

83498

LOINC® Information

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