

Congenital Adrenal Hyperplasia (CAH)

Use

Quantitative determination of human 17 β -OH-progesterone (17-OHP) in blood specimens dried on filter paper as an aid in screening newborns for congenital adrenal hyperplasia (CAH).

Clinical Significance

There are various congenital enzyme defects of steroid biosynthesis that cause CAH. They are genetically different but are all transmitted in an autosomal recessive manner. The most frequent types are 21 β -hydroxylase deficiency (about 80% of all cases) and 11 β -hydroxylase deficiency (about 15% of all cases). CAH due to 21 β -hydroxylase deficiency is characterized by a deficiency in the hormones, cortisol and aldosterone, and an over-production of androgen. Serious loss of body salt and water can result in death. In girls, the genitalia may appear like that of a male and can result in incorrect sex assignment. Symptoms of adrenal insufficiency can include emesis, excessive weight loss relative to birth weight, diaphoresis, hyperventilation, pallor, dry mucosa, and lethargy. 17-OHP, a precursor of cortisol, is increased in both 21 and 11 β -hydroxylase deficiency. Its determination is thus useful as a screening method for the two most frequent types of CAH or about 95% of all cases.

Further information and ACT Sheets can be found at the OSDH Newborn Screening Program [website](#).

Methodology

GSP Neonatal 17 α -OH-progesterone (17-OHP) solid phase, time resolved, fluoroimmunoassay. Cases with elevated 17-OHP are referred for 2nd tier tandem mass spectroscopy (MS/MS) steroid profiling.

Specimen Type

See [Guidance for Collection of NBS Dried Blood Spots](#)

Minimum Volume/Size

See [Guidance for Collection of NBS Dried Blood Spots](#)

Collection Instructions

See [Guidance for Collection of NBS Dried Blood Spots](#)

Common Causes for Rejection

See [Guidance for Collection of NBS Dried Blood Spots](#)

Shipping

See [Guidance for Collection of NBS Dried Blood Spots](#)

Turn-around Time

- Within 5 working days of receipt
- Within 7 working days of receipt if 2nd tier steroid profile is required

Reference Range

Within Normal Limits

- 17-OHP < 28.0 ng/mL if birth weight is \geq 2500 grams
- 17-OHP < 75.0 ng/mL if birth weight is < 2500 grams
- Steroid Profile: Normal

Reportable Results

- 17-OHP
 - Within Normal Limits
 - Outside Normal Limits, <value> ng/mL
- Steroid Profile
 - Normal
 - Abnormal

Interpretation

Primary Screening

- Within Normal Limits
 - Not Consistent with CAH

Following Secondary Steroid Profiling

- Birth weight is \geq 2500 grams
 - Elevated 17-OHP With Normal Steroid Profile: Not consistent with congenital adrenal hyperplasia.
 - Elevated 17-OHP With Abnormal Steroid Profile: Submit repeat specimen as soon as possible.
 - High 17-OHP With Normal Steroid Profile: 17-OHP value outside normal limits with normal steroid profile; submit repeat specimen as soon as possible.
 - High 17-OHP With Abnormal Steroid Profile: Consistent with congenital adrenal hyperplasia. Immediate confirmatory testing recommended.
- Birth weight is < 2500 grams
 - Elevated 17-OHP With Normal Steroid Profile: Not consistent with congenital adrenal hyperplasia.
 - Elevated 17-OHP With Abnormal Steroid Profile: Submit repeat specimen as soon as possible.
 - High 17-OHP With Normal Steroid Profile: 17-OHP value outside normal limits with normal steroid profile. Not consistent with classic congenital adrenal hyperplasia; no follow-up needed unless clinically indicated.
 - High 17-OHP With Abnormal Steroid Profile: Consistent with congenital adrenal hyperplasia. Immediate confirmatory testing recommended.

Limitations/Interferences

- This is a screening test only; a diagnostic procedure should be used to confirm a diagnosis of CAH.
- Late onset, non-classic CAH is not accurately detected by NBS.
- NBS for CAH is not intended to detect mild cases, although some are detected.
- Preterm or low birth weight and samples taken at \leq 24 hours of age may cause false-positive results.
- Treatment of the mother or child with dexamethasone, hydrocortisone, or prednisone may result in false-negative results.
- Specimens improperly collected, processed or transported may result in erroneous results.

CPT Code

83498

Notes

The GSP Neonatal 17 α -OH-progesterone assay is approved for *in vitro* diagnostic use by the U.S. Food and Drug Administration. Second-tier steroid profile testing is performed at Mayo Clinic Laboratories, 200 First St. SW, Rochester, MN 55905 and is not cleared or approved by the U.S. Food and Drug Administration.