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State-Recommended Guidelines for Follow-up of a Positive Newborn Screen for Congenital Adrenal Hyperplasia (CAH)

Babies in the Neonatal Intensive Care Unit

These general guidelines have been developed by a committee of pediatric endocrinologists in collaboration with the California Newborn Screening Program to assist neonatologists in the follow-up of a positive CAH newborn screen until the NICU baby can be evaluated by a pediatric endocrinologist. The California Newborn Screening Program **strongly** recommends that a CCS-approved endocrine center (see attached list of pediatric endocrine centers) or CCS-paneled endocrinologist be contacted for consultation on the diagnostic evaluation and initiation of treatment of these newborns if indicated. The newborn screening coordinator who reported the positive test to you can assist in making a referral. Diagnostic services rendered by a center/specialist are covered by CCS in the absence of insurance.

There is a relatively high rate of false-positive newborn screening results for CAH in NICU babies, reflecting the degree of prematurity and extent of critical illness. However, because about 2/3 of the true positives identified by newborn screening have the potentially life-threatening salt-wasting variant of CAH, there is an urgent need for prompt evaluation.

California law requires that babies with positive screens have confirmatory testing. Repeating the newborn screen is inappropriate unless recommended by a pediatric endocrinologist.

- If the infant has ambiguous genitalia at birth, **immediately refer the baby to or consult with a pediatric endocrinologist, who will order appropriate laboratory tests.** Keep in mind that premature males may normally have undescended testicles and premature females may have a relatively large clitoris. In fact, CAH is only one possible cause of ambiguous genitalia.
 - ❖ If the infant has normal genitalia and the newborn screen is positive, order a confirmatory serum 17-hydroxyprogesterone (17-OHP) immediately. It is recommended that specimens be sent to either Esoterix (1-800-444-9111) or Quest Diagnostics (1-800-553-5445) to confirm or rule out this condition. These facilities require a volume of only 0.1 to 0.2 mL of serum for a 17-OHP and assure a rapid turn-around time of one to two days from the time that the specimen is received at their laboratory. The facility drawing the blood specimen should contact the preferred laboratory to obtain specimen collection and handling instructions, and to alert it to the impending arrival of the specimen. The laboratory should be informed that the patient is a neonate, as well as whom to contact with the result. Tests should be ordered **STAT**. **Refer the baby to or consult with a pediatric endocrinologist.**

Make sure that these test codes are used when ordering serum 17-OHP:

Esoterix: 501847 Neonatal, 17-OHP, STAT
Quest: 17654X 17-Hydroxyprogesterone, Neonatal/Infant

- Closely monitor the baby's condition, observing for vomiting, lethargy, poor feeding, and signs of dehydration and shock.
- Monitor serum sodium and potassium and plasma glucose periodically since abnormalities in these analytes usually are not apparent until 10-20 days of age. If, at any time, there is significant hyponatremia and/or hyperkalemia, with or without hypoglycemia, and/or if there is clinical deterioration, obtain a **STAT** 17-OHP level. Then start IV hydrocortisone at a dose of 100 mg/m²/day (term babies are typically 0.25-0.3 m² and, therefore, the total daily dose is 25 mg for a term infant and proportionately less in a preemie depending on the weight), to be administered either as a continuous infusion (~1 mg/hour) or at intervals (*i.e.*, daily dose divided every 6 hours).
- If the first confirmatory 17-OHP is greater than 3000 ng/dL, consult with a pediatric endocrinologist. If the first confirmatory 17-OHP is equal to or less than 3000 ng/dL and the baby is stable, repeat the test (second confirmatory test) within 3 weeks or prior to discharge, whichever comes first.
- If the second confirmatory 17-OHP result is greater than 1500 ng/dL, consult with a pediatric endocrinologist. If the second confirmatory 17-OHP result is equal to or less than 1500 ng/dL, repeat the test at the time the infant would have been term. At term, normal 17-OHP values are less than 450 ng/dL.
- If the baby has declining levels of 17-OHP over time, CAH can likely be ruled out.

References

Ballerini MG, Chiesa A, Scaglia P, Gruñeiro-Papendieck L, Heinrich JJ, Ropelato MG. 17 alpha-hydroxyprogesterone and cortisol serum levels in neonates and young children: influence of age, gestational age, gender and methodological procedures. *J Pediatr Endocrinol Metab* 2010;23:121-132

Speiser PW, Azziz R, Baskin LS, Ghizzoni L, Hensle TW, Merke DP, Meyer-Bahlberg HF, Miller WL, Montori VM, Oberfield SE, Ritzen M, White PC: Endocrine Society. Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency; an Endocrine Society clinical practice guideline. *J Clin Endocrinol Metab* 2010;95:4133-4160.