



KAREN L. SMITH, MD, MPH  
Director and State Health Officer

State of California—Health and Human Services Agency  
California Department of Public Health



EDMUND G. BROWN JR.  
Governor

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

### Newborns Who Are Home When Screening Results Return

These general guidelines have been developed by a committee of pediatric endocrinologists in collaboration with the California Newborn Screening (NBS) Program to assist primary care providers in following up on a positive CAH screen until the baby can be seen by an endocrinologist. The NBS Program **strongly** recommends that babies with a positive newborn screen for CAH be referred to a CCS-approved pediatric endocrinology center (see attached list) or a CCS-paneled pediatric endocrinologist for diagnostic evaluation and initiation of treatment when indicated. Diagnostic services provided by a center/specialist are covered by CCS in the absence of insurance. A coordinator from the Newborn Screening Area Service Center (ASC) that reported the positive result to you will facilitate the referral. There is a significant likelihood that a term newborn with a positive newborn screen for CAH has the disorder. For this reason and because CAH is potentially life-threatening, the diagnostic evaluation should begin immediately.

**1. Have the baby admitted to a hospital immediately (or send the baby to an emergency department) for diagnostic evaluation and treatment.**

- Order the following tests to be run **STAT**: serum 17-hydroxyprogesterone (17-OHP), glucose, and electrolytes.
- Serum testosterone and plasma renin activity (PRA) should also be ordered.
- It is recommended that all hormone testing be performed at either Esoterix (800-444-9111) or Quest Diagnostics (800-553-5445), both of which can assure a 17-OHP result within one to two days from the time the specimen arrives at the 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.

Make sure that these test codes are used when ordering the tests:

Esoterix:

17-OHP: 501847 (Neonatal, 17-OHP STAT)  
PRA: 500278

Quest:

17-OHP: 17654X (17-Hydroxyprogesterone, Neonatal/Infant)  
PRA: 16846

- The admitting physician/neonatologist should call the laboratory in 24-48 hours to obtain the result if the laboratory has not phoned him or her.
2. Refer the baby to a pediatric endocrinologist within 24 hours for diagnostic evaluation and management. The ASC coordinator will contact the endocrine center/specialist of your choice and facilitate CCS coverage. If the specialist cannot see the baby, he/she should be consulted on patient management.
  3. If a pediatric endocrinologist cannot be reached, start treatment immediately after the confirmatory blood tests have been drawn. Do not withhold treatment until test results are reported unless advised to do so by a pediatric endocrinologist.
    - For critically ill infants:
      - Start IV hydrocortisone at a dose of 100 mg/m<sup>2</sup>/day. Babies at term are typically 0.25 to 0.3 m<sup>2</sup> and, therefore, the total **daily** dose is 25-30 mg to be administered either as a continuous infusion (~1 mg/hour) or at intervals (*i.e.*, daily dose divided, to be given every 6 hours).
    - For clinically stable infants:
      - Treatment should be initiated with oral administration of:
        - 1) Hydrocortisone tablets (5-mg strength) using either generic hydrocortisone or Cortef® brand at a dose of 5 mg (1 tab) three times per day is preferred while the compounded suspension (in a concentration of 10 mg/5 mL = 2 mg/mL) is not to be used unless recommended by your pediatric endocrinologist.
        - 2) Fludrocortisone (0.1-mg strength) at 0.1 mg (1 tab) per day, with subsequent adjustments to be made by the pediatric endocrinologist.
        - 3) Tablets should be crushed, mixed with less than one teaspoon of breast milk, formula, or water, and given by spoon or syringe.
      - If the 17-OHP is normal, do not discontinue hydrocortisone abruptly if it has been administered for more than one week. Discuss the taper procedure with the endocrinologist.

Do not discharge the baby until the blood test results are normal or the baby is on treatment, with test results pending.

    - Consult with the pediatric endocrinologist prior to or within 24 hours of discharge.
    - Note the test results in the medical record.

#### References:

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.

Ballerini MG, Chiesa A, Scaglia P, Gruñiero-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.

July 2012

California Department of Public Health, Newborn Screening Program with the Pediatric Endocrine Guidelines Committee