Provider Action Sheet for Congenital Adrenal Hyperplasia (CAH)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for congenital adrenal hyperplasia (CAH), which is TIME-CRITICAL and requires immediate follow-up.

What steps do I need to take?

1. **Consult today** with a California Children's Services Endocrine Special Care Center (SCC) specialist for guidance. They will ask for your assessment of the infant's current health status and if the infant has any symptoms or signs of CAH. Further evaluation and confirmatory testing may be required, including hospital admission. If the infant is diagnosed with CAH, the SCC will provide follow-up clinical care.
2. **Contact** the family to explain the positive newborn screening test result. For information on how best to communicate this, consult the **Health Resources & Services Administration Newborn Communication Guide** (https://bit.ly/HRSAGuide or search online for "hrsa" "heritable" "communication").
3. **Review** the accompanying "Family Action Sheet for Congenital Adrenal Hyperplasia (CAH)" with the family and ensure they understand it. Educate them about the symptoms and signs of adrenal crisis and the need for immediate medical care if these occur.
4. **Inform** the family that this result requires more testing and evaluation. Emphasize that you, along with NBS Program Area Service Center (ASC) staff and the SCC Endocrine specialist, will guide them through the next steps of confirmatory testing and follow-up services.
5. **Advise** parents to (1) follow the plan for confirmatory testing right away, and (2) keep their infant's appointments with an SCC specialist, if any are scheduled. The family should receive services from a multidisciplinary team of specialists.

Clinical information

CAH encompasses a group of autosomal recessive disorders where there is deficiency of an enzyme required for adrenal cortisol synthesis. The most common form is 21-Hydroxylase Deficiency (21-OHD). Severe enzyme deficiency leads to **classic CAH,** which occurs in the **simple virilizing form** (~25% of cases) or the **salt-wasting form** (~75% of cases), which poses risk for a life-threatening salt-wasting crisis.

Infants with CAH may present with poor feeding, vomiting, dehydration, hypotension, and lethargy. Biological females with CAH may exhibit ambiguous genitalia. The primary treatment for CAH is corticosteroid replacement, which may be lifelong. Surgery may be considered for females with ambiguous genitalia

Milder enzyme deficiencies lead to **non-classic CAH**, which is less severe but may require treatment if symptomatic.

Please refer to the site below for additional clinical information.

ACMG ACT Sheets and Algorithms

(www.ncbi.nlm.nih.gov/books/NBK55827/)

Questions?

For follow-up questions, please call your NBS Program Area Service Center.

For program questions, please email NBS Program staff at NBS@cdph.ca.gov or visit the NBS Program website (www.cdph.ca.gov/NBS).

California Department of Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)