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):

What steps do I need to take?

1. **Review** the California state recommended guidelines for follow up on CAH provided by the Area Service Center (ASC) staff. **Confirmatory testing is needed immediately.**
2. **Consult** with a California Children's Services Special Care Center (SCC) endocrinology specialist. They will ask for your assessment of the infant's current health status and whether the infant has any signs of the disorder as they help formulate a diagnostic follow-up plan. Further evaluation may be required to make a final diagnosis. If the infant has a diagnosed condition, the SCC will provide clinical follow-up care.
3. **Contact** the family to explain the positive newborn screening test result. For information on how best to communicate, consult the **Health Resources & Services Administration Newborn Communication Guide** (https://bit.ly/HRSAGuide or search online for "hrsa" "heritable" "communication").
4. **Review** the accompanying "Family action sheet for congenital adrenal hyperplasia (CAH)." with the family and ensure they understand.
5. **Remind** the family that their infant was positive on a screen, and it is not diagnostic. Emphasize that you, with the help of ASC staff and the SCC endocrinology specialist, will guide them through the next steps of confirmatory testing and follow-up services.
6. **Advise** parents to (1) follow the plan for confirmatory testing right away, and (2) keep their infant's appointments with an SCC endocrinology specialist, if any are scheduled. The family should receive services from a multidisciplinary team of specialists.

Clinical information

CAH is a group of disorders characterized by the deficiency of an enzyme required for the synthesis of cortisol in the adrenal glands. The most common form is 21-Hydroxylase Deficiency. Classical CAH can present as salt-wasting and/or simple-virilizing forms.

CAH makes infants susceptible to poor feeding, vomiting, dehydration, hypotension, and lethargy. The primary treatment of CAH is corticosteroid replacement as needed. Surgery for females with ambiguous genitalia may be considered.

Please visit these sites. Search for the site name and "congenital adrenal hyperplasia" if needed.

MedlinePlus
https://medlineplus.gov/ency/article/000411.htm

National Institutes of Health
https://www.nichd.nih.gov/health/topics/cah#

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)