ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet

[Elevated 17-hydroxyprogesterone (17-OHP)]

Congenital Adrenal Hyperplasia (CAH)

Differential Diagnosis: Classic Congenital Adrenal Hyperplasia (CAH) caused by 21-hydroxylase deficiency; Non-classic 21-hydroxylase deficiency caused by 21-hydroxylase deficiency; 11 beta-hydroxylase deficiency; prematurity.

Condition Description: CAH is caused by a deficiency of one of the enzymes required to synthesize cortisol in the adrenal glands. The most common type is 21-hydroxylase deficiency, resulting in impaired aldosterone and cortisol synthesis. 21-hydroxylase deficiency has two major forms: the salt wasting form, which can lead to adrenal crises, and the simple virilizing form. The simple virilizing form can cause virilization in biologic females. Non-classic 21-hydroxylase deficiency can cause hyperandrogenism and virilization later in childhood; it does not cause salt wasting adrenal crises, and may not be detected by newborn screening. 11- beta-hydroxylase deficiency may present in the newborn periodwith 17-OHP elevations.

You Should Take the Following IMMEDIATE Actions If The Infant Is Not Premature:*

- Inform family of the newborn screening result.
- Ascertain clinical status (lethargy, vomiting, poor feeding).
- Consult with pediatric endocrinologist the same day.
- Immediately evaluate the newborn (lethargy, vomiting, ambiguous genitalia) and obtain serum electrolytes and glucose .
- Immediately transport to a hospital for further treatment in consultation with a pediatric endocrinologist.
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide the family with basic information about CAH. Educate the family about signs and symptoms of adrenal crises, including the need for urgent treatment.

- Report final diagnostic outcome to the state newborn screening program.

Diagnostic Evaluation: Adrenal steroid hormone panels: confirm the diagnosis. ACTH stimulation testing, electrolyte changes with hyponatremia/hyperkalemia, and increased renin : support the diagnosis. Molecular genetic testing : may be used to support a diagnosis of CAH and may help to determine the type of 21-hydroxylase deficiency.

Clinical Considerations: Newborns with the classic form of CAH are at risk for adrenal crises with illness, surgery, or trauma resulting in shock and death if untreated. Ambiguous genitalia can occur in biological females with CAH. A neonate with CAH may require lifelong adrenal hormone replacement therapy and often requires the care of a multidisciplinary team; therapy depends on the form of CAH.
**Premature infants have an increased rate of false positive test results; recommendations for 17-OHP follow up may be different for premature infants.*

Additional Information:

- How to Communicate Newborn Screening Results
- Gene Reviews
- Medline Plus
- Pediatric Endocrine Society
- Endocrine Society Congenital Adrenal Hyperplasia Guideline Resources
- Clinicaltrials.gov

Referral (local, state, regional, and national):

- Find a Genetics Clinic Directory
- Genetic Testing Registry
- Find a Pediatric Endocrinologist
- Cares Foundation

 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks*)*

Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results
- https://bit.ly/NBSResultsHRSA

Gene Reviews
- https://www.ncbi.nlm.nih.gov/books/NBK1171/

Medline Plus
- https://medlineplus.gov/genetics/condition/21-hydroxylase-deficiency/

Clinicaltrials.gov
- https://clinicaltrials.gov/

Pediatric Endocrine Society
- https://pedsendo.org/patient-resource/congenital-adrenal-hyperplasia/

Endocrine Society Congenital Adrenal Hyperplasia Guideline Resources
- https://www.endocrine.org/clinical-practice-guidelines/congenital-adrenal-hyperplasia-guideline-resources

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
- https://clinics.acmg.net

Genetic Testing Registry
- https://www.ncbi.nlm.nih.gov/gtr/

Find a Pediatric Endocrinologist
- https://pedsendo.org/patient-resources/find-a-pediatric-endocrinologist/

Cares Foundation
- https://caresfoundation.org/

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