

Newborn Screening ACT Sheet

[Elevated 17-hydroxyprogesterone (17-OHP)]

Congenital Adrenal Hyperplasia (CAH)

Differential Diagnosis: Congenital Adrenal Hyperplasia (CAH), 21-OH deficiency; stress or prematurity are possible secondary causes of increased 17-OHP.

Condition Description: Lack of adequate adrenal cortisol and aldosterone, and increased androgen production.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
 - Consult with pediatric endocrinologist, having the following information available (sex, age at NBS sampling, birth weight) and refer, if needed.
 - Examine the newborn (ambiguous genitalia or non palpable testes, lethargy, vomiting, poor feeding).
 - Initiate timely confirmatory/diagnostic testing as recommended by specialist.
 - Emergency treatment as indicated (e.g. IV fluids, IM/IV hydrocortisone).
 - Educate family about signs, symptoms and need for urgent treatment of adrenal crisis.
 - Report findings to newborn screening program.
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Diagnostic Evaluation: Diagnostic tests include serum 17-OHP (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced). Additional tests may be recommended by the specialist.

Clinical Considerations: Ambiguous genitalia in females who may appear to be male with non-palpable testes. Infants with Congenital Adrenal Hyperplasia are at risk for life-threatening adrenal crises, shock, and death in males and females. Finding could also be a false positive associated with stress or prematurity.

Additional Information:

[Gene Reviews](#)

[Cares Foundation](#)

[Genetics Home Reference](#)

[ClinGen Actionability Report](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Lawson Wilkins Pediatric Endocrine Society "Find A Doc"](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site *(insert local and regional newborn screening website information)*

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews

<http://www.ncbi.nlm.nih.gov/books/n/gene/cah>

Cares Foundation

<http://caresfoundation.org>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=21hydroxylasedeficiency>

ClinGen Actionability Report

<https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC151>

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2192?db=genetests

Clinical Services

Lawson Wilkins Pediatric Endocrine Society “Find A Doc”

<http://lwpes.org/>

Find Genetic Services

<https://clinics.acmg.net>

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