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.

Diagnostic Evaluation: Diagnostic tests include serum 17-0HP (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

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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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