



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.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- 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-OHP (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced)**. Additional tests may be recommended by the specialist.

Clinical Expectations: Ambiguous genitalia in females who may appear to be male with nonpalpable testes. 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:

(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

[Gene Tests/Gene Clinics](#)

[Cares Foundation](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

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

Contact local/regional University-affiliated medical center

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.

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

Name
URL
Comments

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

Name
URL
Comments

Name
URL
Comments

Name
URL
Comments

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Tests/Gene Clinics

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=PDqbsAPHeqXyI&gry=&fcn=y&fw=mhZr&filename=/profiles/cah/index.html>

Cares Foundation <http://caresfoundation.org>

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

Referral (local, state, regional and national):

Lawson Wilkins Pediatric Endocrine Society “Find A Doc” <http://lwpes.org/>

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