



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.
- Initial testing: serum electrolytes, 17-OHP.
- Repeat the newborn screen if the second screen has not been done.
- 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 non-palpable 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:

OMIM

<http://www.omim.org/entry/201910?search=cah&highlight=cah>

Cares Foundation

<http://caresfoundation.org>

Genetics Home Reference

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