



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.

Please Take the Following Immediate Actions:

- **Immediate phone consult with pediatric endocrinologist**, initiate timely confirmatory/diagnostic testing as recommended by the specialist. (See attached list)
- **Contact family immediately** to inform them of the newborn screening result and ascertain clinical status.
- **Examine the newborn immediately** (Assess for ambiguous genitalia or non-palpable testes, lethargy, vomiting, diarrhea, dehydration, poor feeding)
- **Educate family** about signs, symptoms, and the need for urgent treatment of adrenal crisis.
- **Immediate initial lab testing: 17-HYDROXYPROGESTERONE and DAILY sodium and potassium.**
- **Emergency treatment** as indicated (e.g., IV fluids, IM/IV hydrocortisone).
- **Collect repeat screen (between 7-14 days of life)** if the second screen has not been done.

Note: a NORMAL repeat screen will NOT clear this case

- **Report findings** to the newborn screening program. **FAX to 512-465-4958**

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

Diagnostic Evaluation: Diagnostic tests include serum 17-HYDROXYPROGESTERONE (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:

How to Communicate Newborn Screening Results

<https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/resources/achdnc-communication-guide-newborn.pdf>

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

Cares Foundation <http://caresfoundation.org>

MedlinePlus <https://medlineplus.gov/genetics/condition/21-hydroxylase-deficiency/>

