TEXAS Health and Human Services
Texas Department of State Health Services
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Commissioner

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 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 need for urgent treatment of adrenal crisis.
- ☐ **Immediate initial 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 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/achdnccommunication-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/