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

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 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;
- **Consult with pediatric endocrinologist**, initiate timely confirmatory/diagnostic testing as recommended by specialist;
- **Initial testing: 17-HYDROXYPROGESTERONE and daily sodium and potassium;**
- **Repeat the newborn screen** if the second screen has not been done;
- **Emergency treatment** as indicated (e.g., IV fluids, IM/IV hydrocortisone); and
- **Report findings** to newborn screening program. – **FAX to 512-776-7421.**

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 Considerations

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

Online Mendelian Inheritance in Man – Entry #201910
Cares Foundation
U.S. National Library of Medicine, Medline Plus – 21-hydroxylase deficiency

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