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.

(See flowchart for information concerning the specific actions)

Diagnostic Evaluation:

Diagnostic tests include serum 17-0HP (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.

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. 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.
**Congenital Adrenal Hyperplasia (Elevated 17-OHP)**

**Newborn Screen Elevated 17-OHP**

- **Mild elevation**
  - Clinical suspicion: Low
    - Repeat newborn screen
  - Normal
    - No further action
  - High
    - Follow actions for moderate to severe elevation

- **Moderate to severe elevation**
  - Clinical suspicion: Low
    - Serum 17-OHP, 'Lytes, glucose
    - Moderately high 17-OHP; NL 'Lytes, glucose
  - Clinical suspicion: High
    - Confirmatory tests for 21-OHD: ACTH, stim, steroid profile, genotype
    - Normal
      - No further action
    - Nonclassic 21-OHD
      - Discretionary treatment
    - Classic 21-OHD
      - Replacement therapy
    - Other enzyme defect
      - Replacement therapy
    - Severely high 17-OHP; Low Na, Replacement therapy High K, Low glucose
      - Replacement therapy
Actions are shown in shaded boxes; results are in the unshaded boxes.

**Abbreviations/Key**

- 17-OHP = 17-hydroxyprogesterone
- 'Lytes = Serum electrolytes
- ACTH stim = Adrenocorticotropic hormone stimulation test
- 21-OHD = 21-hydroxylase deficiency
- Steroid profile = Complete adrenal cortical hormone profile, e.g., by MS/MS
- Discretionary treatment = Consult pediatric endocrinology to determine if hydrocortisone therapy is necessary

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