



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED 17-HYDROXYPROGESTERONE (17-OHP)

CONDITION: CONGENITAL ADRENAL HYPERPLASIA (CAH)

DIFFERENTIAL DIAGNOSIS: Congenital Adrenal Hyperplasia (CAH), 21-OH deficiency; stress or prematurity are possible secondary causes of increased 17-OHP.

METABOLIC DESCRIPTION: Lack of adequate adrenal cortisol and aldosterone, and increased androgen production.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- ◆ Contact the family to inform them of the newborn screening result and ascertain clinical status.
- ◆ Examine the newborn (ambiguous genitalia or non-palpable testes, lethargy, vomiting, poor feeding).
- ◆ If the 17-OHP level is Borderline Risk and the baby has no clinical indicators, the Newborn Screening should be repeated prior to confirmatory testing.
- ◆ If the 17-OHP level is High Risk, consult with pediatric endocrinologist, having the following information available (sex, age at NBS sampling, birth weight) and refer, if needed.
- ◆ 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.

CONFIRMATION OF DIAGNOSIS: 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. Infant is 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.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

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KU Medical Center
Office: 913-588-6326

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Wichita Endocrinology
Office: 316-777-6404

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DISCLAIMER: These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. 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. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.