

**Physician ACT SHEET for POSITIVE Newborn Screening Result for CONGENITAL ADRENAL HYPERPLASIA**

- **Consult with Pediatric endocrinologist**, having the following information available (sex, age at NBS sampling, birth weight) and refer, if needed.
- **IMMEDIATELY examine the newborn** (ambiguous genitalia or non palpable testes), **Assess for salt-wasting symptoms** - (emesis, diaphoresis, pallor, lethargy, excessive weight loss relative to birth weight, hyperventilation, dry mucosa).
- **Initial lab work (Do not administer any medications until all specimens have been collected): electrolytes (locally), steroid profile (at reputable laboratory)**
- **IV fluid (D10 0.5NS) replacement if dehydrated.**
- **Admit patient to hospital for observation** due to failed NBS (ICD-9 796.6). Even if minimal or no symptoms, monitoring is critical.
- **Consider administration of IV or IM Solu-Cortef 50mg/M<sup>2</sup> AFTER obtaining steroid profile**

**Disease Category:** Adrenal endocrinopathy

**MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:**

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**Pediatric Endocrinology contacts:** (402) 955-3871

**Meaning of the Screening Result:** Congenital Adrenal Hyperplasia (CAH), 21-OH deficiency; stress or immaturity are also possible causes for increase.

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

**Confirmation of Diagnosis:** Diagnostic tests include serum 17-OHP (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced). Additional tests such as specific steroid profiles may be recommended by specialist.

**Clinical Expectations:** Ambiguous genitalia in females. Infant may appear to be male with nonpalpable testes. Males will appear normal. Feeding problems, life-threatening adrenal crisis, shock, and **death** in males and females.