Disease Name	**Congenital Adrenal Hyperplasia**
Acronym	CAH
Disease Classification	Endocrine Disorder
Symptom onset	INFANTS WITH CAH DO NOT APPEAR ILL AT BIRTH, BUT MAY, WITHIN THE FIRST FEW WEEKS OF LIFE, EXPERIENCE A SALT-LOSING CRISIS WHICH CAN LEAD TO SERIOUS ILLNESS AND DEATH.
Symptoms	Congenital adrenal hyperplasia (CAH) results from a deficiency in one or another of the enzymes of cortisol biosynthesis. In about 95% of cases, 21-hydroxylation is impaired in the zona fasciculata of the adrenal cortex so that 17-hydroxyprogesterone (17-OHP) is not converted to 11-deoxycortisol. Because of defective cortisol synthesis, ACTH levels increase, resulting in overproduction and accumulation of cortisol precursors, particularly 17-OHP, proximal to the block. This causes excessive production of androgens, resulting in virilization.
Natural history without treatment	If untreated, children with CAH will experience abnormally rapid growth early in childhood (but stunted in the long run) and early appearance of body hair. Babies with the salt-wasting form of CAH (about 75 percent of cases) are at risk for rapid, uncontrolled loss of salt from the body that can result in death. The imbalance of hormones before birth may cause some girls to have ambiguous genitalia.
Treatment	Daily supplements of the hormone cortisol, and in many cases a salt-retaining hormone. To prevent problems, treatment must begin shortly after birth.
Physical phenotype	Ambiguous genitalia in females
Inheritance	Autosomal recessive
General population incidence	1 in 21,500
OMIM Link	http://www.ncbi.nlm.nih.gov/omim/201910
Genetests Link	www.genetests.org
Support Group	Congenital Adrenal Hyperplasia Research Education & Support http://www.caresfoundation.org MAGIC Foundation for Children's growth (MAGIC) http://www.magicfoundation.org National Organization for Rare Diseases http://www.rarediseases.org

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.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- 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.

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 Considerations: Ambiguous genitalia in females who may appear to be male with non-palpable testes. Infants with Congenital Adrenal Hyperplasia are 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:

Gene Reviews
Cares Foundation
Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services
Lawson Wilkins Pediatric Endocrine Society "Find A Doc"