



## 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.
- Initial testing: serum electrolytes, 17-OHP.
- Repeat the newborn screen if the second screen has not been done.
- 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-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. 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 Tests/Gene Clinics

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=PDqbsAPHeqXyl&gry=&fcn=y&fw=mhZr&filena me=/profiles/cah/index.html>

##### Cares Foundation

<http://caresfoundation.org>

##### Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=21hydroxylasedeficiency>



## Newborn Screening FACT Sheet

### Congenital Adrenal Hyperplasia (CAH)

#### What is CAH?

Babies with Congenital Adrenal Hyperplasia (CAH) are born with enlarged adrenal glands. The adrenal glands of these babies can't make enough of a hormone called cortisol — the main hormone in adrenal glands. The adrenal glands get too big when they try to make the right amount of this hormone.

#### What Causes CAH?

The adrenal glands make chemical messengers called hormones. The hormones they make are cortisol (hydrocortisone), aldosterone (salt-retaining hormone), and androgens (male sex hormones). CAH happens when certain enzymes in the adrenal glands are missing. Enzymes start chemical reactions in the body. The missing enzymes cause the glands to make too little of the cortisol and aldosterone hormones and too much of the male-like hormones. CAH is an inherited disorder. Both parents carry the gene for CAH.

#### What Symptoms or Problems Occur with CAH?

*[Symptoms are something out of the ordinary that a parent notices.]*

There are three main forms of CAH:

- severe salt-wasting
- non-salt wasting
- milder form

An infant with the severe salt-wasting form may have one or more of these symptoms in the first weeks of life:

- vomiting
- poor weight gain
- poor feeding
- sleepiness
- diarrhea
- dehydration (loss of fluids)

Newborns with the non-salt wasting form of CAH usually don't get severely sick. A girl's genitals may look more like a boy's. Other symptoms of

the non-salt wasting form develop with age. Both boys and girls may have:

- rapid growth in early childhood
- early sexual development
- early pubic hair growth

Children with the milder form of CAH show symptoms anytime between early childhood and puberty. They might grow more quickly or grow pubic hair early. Girls at puberty usually have:

- excess body hair growth
- acne
- irregular periods
- sometimes infertility

#### What is the Treatment for CAH?

**Medications** – CAH is treated with two types of hormone medicines. Both boys and girls with CAH need to take a cortisol-like hormone pill. This will prevent the body from making too much of the male-like hormone. Children with CAH take the pills two or three times a day. Children with salt-wasting CAH may also take a salt-saving hormone pill, plus salt tablets. Your doctor may tell you to add salt to your baby's formula. A cortisol shot may be necessary if your child gets sick.

#### Things to Remember

Children with CAH must take hormone pills all of their lives. Your doctor will check your child's height, weight, and blood pressure. He or she may x-ray the wrist to look at the bone age and may do a blood test to check hormone levels. Your doctor will prescribe the hormone medicine after testing. He or she may change the amount as your child grows or gets sick or hurt.