



Newborn Screening FACT Sheet

Congenital Adrenal Hyperplasia (CAH)

What is CAH?

People with Congenital Adrenal Hyperplasia (CAH) have problems with hormone production. These hormones help regulate glucose, salt, and sex characteristics in the body. This can lead to low glucose levels, low sodium levels, and atypical genital formation.

What Causes CAH?

The adrenal gland hormones are cortisol that helps with glucose in the body, aldosterone that helps with salt retention, and androgens that cause male sex characteristics. CAH happens when the adrenal glands do not function correctly. CAH is an inherited disorder. Both parents carry the gene for CAH.

What Symptoms or Problems Occur with CAH?

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. Female genitals may appear more like a male's. A male with CAH

may not have any effects on their genitals. Other symptoms of the non-salt wasting form develop with age. Both boys and girls may have:

- rapid growth in early childhood
- early onset puberty changes
- early pubic hair growth

Children with the milder form of CAH could 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 hormone replacement. This will prevent the body from making too much androgen. Children with salt-wasting CAH may also take a salt-saving hormone replacement, plus salt replacement tablets. If your child becomes ill, from a cold or virus, they may need extra medication to help their body with the stress of the illness.

Things to Remember

Children with CAH must take hormone replacements all of their lives. Your doctor will do a blood test to check hormone levels. If needed, your doctor will prescribe any medicine after testing and confirming the diagnosis. This is a lifetime condition that will require monitoring by an endocrinologist.

Resources:

Baby's First Test

<https://babysfirsttest.org/newborn-screening/conditions/congenital-adrenal-hyperplasia>

Navigate Life Texas <https://www.navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed>

