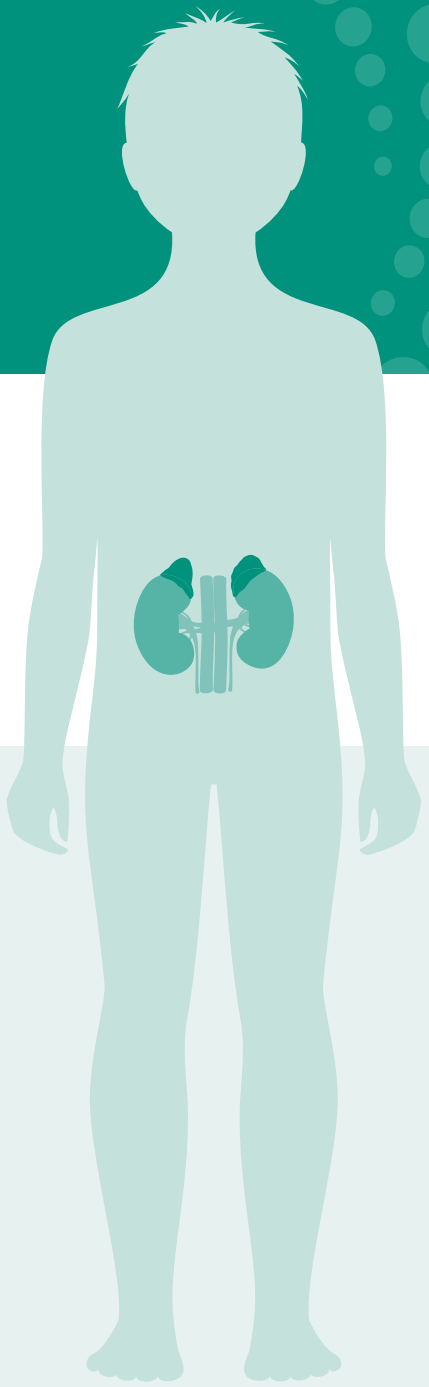
THE TRUTH ABOUT CONGENITAL ADRENAL HYPERPLASIA (CAH)

Congenital Adrenal Hyperplasia, also referred to as CAH, is a genetic disorder in which the two adrenal glands do not function properly because of mutations in the gene encoding adrenal steroid 21-hydroxylase, which is an enzyme. Without this enzyme, the adrenal glands may produce too little cortisol and/or aldosterone and too much androgen. Children can inherit an abnormal copy of the gene from each of their parents.



HOW IS CAH DIAGNOSED?

Newborns are screened for CAH with a blood test from a heel prick right after birth, which is known as newborn screening. Diagnosis in infancy or later may also include:

- A history and physical examination
- Blood tests
- Genetic test

WHAT ARE THE DIFFERENT TYPES OF CAH?

CAH is a type of inherited disorder called “autosomal recessive.” Meaning a disorder that can be passed from parents to their children. For a child to have CAH, each parent must either have CAH or carry an abnormal gene. There are two major types, CAH disorder can be severe (classic) or mild (non-classic).

CLASSIC CAH

Classic CAH is usually diagnosed during infancy or early childhood, it is the most severe. In one form of classic CAH, called “salt-wasting” (meaning the body has trouble keeping the right amount of salt in the blood), the adrenal glands do not make enough of the hormones cortisol and aldosterone. If left untreated, classic CAH can cause shock, coma, and death. Another form of classic-CAH is “non salt-wasting”, when is the loss of enzyme activity is less severe. Signs and symptoms include:

- Ambiguous genitalia (external sex organs that resemble male genitals) in female infants
- Rapid growth and early puberty in male children
- Weight loss, frequent vomiting, dehydration, diarrhea, changes in body chemistry, shock, and coma may occur if not diagnosed during infancy

Individuals diagnosed with CAH should have a health care team which includes: a pediatric endocrinologist, a pediatric urology specialist, a mental health expert, and a genetics specialist.

Treatment of Classic CAH ensures proper hormone levels and promotes hormone growth, as well as improving sexual development and sexual function.Glucocorticoids are used as medication to replace the cortisol that patients with classic CAH cannot make. Patients with classic CAH, especially those with the salt-wasting form, also need medicines called mineralocorticoids to replace the aldosterone they can’t make. Newborns also may need salt supplements.

Visit hormone.org for more information.

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NON-CLASSIC CAH

Unlike classic CAH, the non-classic form is mild and not life threatening. Signs and symptoms may or may not appear until childhood or adulthood.

Symptoms in males and females include:

- Early development of armpit and pubic hair
- Early or severe acne
- Infertility or decreased fertility

Adolescent girls and adult women also may have:

- Masculine characteristics such as facial hair and a deep voice
- Infrequent or absent menstrual periods

Some patients with non-classic CAH may have no symptoms and may not require treatment. With proper care, people with either type of CAH can live long and healthy lives.

WHEN TO SEE A DOCTOR:

- If there is a known family history of CAH, a diagnosis can be made before birth.
- If you're pregnant and have a medical history of CAH.
- If you have concerns about your child's growth or development, make an appointment with your child's pediatrician.
- If you have impaired fertility see a reproductive endocrinologist and/or fertility specialist.

QUESTIONS TO ASK YOUR HEALTHCARE PROVIDER:

- What kind of CAH does my child (or do I) have?
- What are the options for treatment for myself or my child?
- What are the risks and benefits of each of the treatment options?
- Should I see a pediatric endocrinologist or any other specialist?
- Should I (or my child) have genetic testing?

Patients have questions. We have answers.

The Hormone Health Network is your trusted source for endocrine patient education. Our free, online resources are available at **hormone.org**.

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