



# Congenital Adrenal Hyperplasia

## What is congenital adrenal hyperplasia (CAH)?

*Congenital* (con-JEN-ih-tuhl) *adrenal* (uh-DREEN-uhl) *hyperplasia* (HY-per-PLAY-zhee-uh), also called CAH, is a group of genetic disorders in which the two adrenal glands do not work properly. Children inherit one gene that causes this disorder from each of their parents. The adrenal glands, located on top of each kidney, make hormones that are essential for body functions.

People with CAH lack one of the enzymes needed for proper function of the adrenal glands. (An enzyme is a protein that causes a chemical change in the body.) Without the enzyme, the adrenal glands may produce too little of the hormones cortisol and/or aldosterone and too much androgen. CAH can be severe (classic) or mild (nonclassic).

## How is CAH diagnosed?

Some forms of CAH are diagnosed at birth. However, milder forms might not be diagnosed until childhood or adulthood. In the United States and many other countries, newborns are checked for CAH with a blood test. Diagnosis in infancy or later also may include

- Further blood tests
- Urine tests
- Genetic tests
- A physical examination
- A family history

Sometimes, when there is a known family history of CAH, a fetus is diagnosed and treated before birth.

## What are the different types of CAH?

There are two types of CAH—classic, which can be life threatening, and non-classic, a milder form of the disorder.

### Classic CAH

Classic CAH, usually first found in infancy or early childhood, is the most severe type of CAH. 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 cortisol and aldosterone. If not found and treated, classic CAH can cause shock, coma, and death. In another form of classic CAH, called “non-salt wasting,” the enzyme shortage is less severe. The adrenal glands make enough aldosterone but not enough cortisol.

### Signs and Symptoms

In many cases, female infants are diagnosed at birth because they have ambiguous genitalia (external sex organs that resemble male genitals). However, they still have normal internal female organs (ovaries and uterus).

A male infant with classic CAH usually appears normal at birth, although he may have an enlarged penis. After infancy, boys with classic CAH grow rapidly and show signs of early puberty.

If infants are not diagnosed at birth, several weeks later they may show weight loss, dehydration, diarrhea, and heart problems. They also may vomit frequently.

### Treatment

The goals of treatment are to ensure proper hormone levels and promote normal growth and sexual development. Patients with classic CAH should have a team of health care providers, including specialists in pediatric endocrinology, pediatric urologic surgery, psychology, and genetics.

People with classic CAH need medicines called glucocorticoids to replace the cortisol their bodies can't make. Extra glucocorticoids may be needed during times

of stress, such as when a patient has an infection. People with classic CAH, especially those with the salt-wasting form, also need medicines called mineralocorticoids. Newborns also may need sodium chloride (salt) supplements.

Surgery can correct ambiguous genitalia in girls. Experts recommend that surgery be performed when the infant is 2 to 6 months of age.

### Nonclassic CAH

Unlike classic CAH, nonclassic CAH is mild and not life threatening. Signs and symptoms might not appear until childhood or adulthood.

### Signs and Symptoms

Signs and symptoms in both males and females include:

- Early development of armpit and pubic hair
- Rapid growth during childhood
- 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

### Treatment

Some patients have no symptoms and require no treatment. Others need low-dose glucocorticoids, but might not need life-long treatment.

## What does the future hold for people with CAH?

With proper care, people with either type of CAH can live long and healthy lives. In the meantime, researchers continue to explore better ways to diagnose and treat this condition.

### Resources

Find-an-Endocrinologist: [www.hormone.org](http://www.hormone.org) or call 1-800-HORMONE (1-800-467-6663)

AboutKidsHealth website from The Hospital for Sick Children (search on CAH): [www.aboutkidshealth.ca](http://www.aboutkidshealth.ca)

National Adrenal Diseases Foundation: [www.NADF.us](http://www.NADF.us) or call 516-487-4992

The CARES Foundation: [www.caresfoundation.org](http://www.caresfoundation.org)

## Some Adrenal Hormones and What They Do

<b>Androgens</b> (male sex hormones)	Regulate growth and male sexual characteristics.
<b>Aldosterone</b> (a mineralocorticoid)	Helps keep a proper balance of salt (sodium) and water in the body. Regulates blood volume and blood pressure.
<b>Cortisol</b> (a glucocorticoid)	Helps the body cope with stress, illness, and injury. Regulates blood glucose and blood pressure levels.

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