

# Congenital Adrenal Hyperplasia: What You Need to Know

**Congenital adrenal hyperplasia (CAH) is a genetic condition that affects the adrenal glands. In this handout, you will learn about CAH, including its symptoms, diagnosis, and treatment.**

## WHAT IS CONGENITAL ADRENAL HYPERPLASIA (CAH)?

**Congenital adrenal hyperplasia (CAH)** is an inherited (genetic) condition that affects the **adrenal glands**. The **adrenal glands** are small glands on top of the kidneys. They make the following important hormones:

- **Cortisol** is the hormone your body makes under stress. It is important for regulating energy and maintaining normal blood pressure and blood sugar. Our bodies make high amounts of cortisol to help us deal with the stress of illness.
- **Aldosterone** helps balance the levels of salt and water in the body. It also helps maintain blood pressure.
- **Androgens** are sex hormones that help with pubertal changes and certain male physical characteristics. Both boys and girls make androgens.

## HOW DOES CAH AFFECT THE BODY?

In CAH, the adrenal glands are missing part of or all an enzyme (typically **21-hydroxylase**). This enzyme helps make cortisol. When almost no enzyme is available, the adrenal glands might not make enough aldosterone as well. Because of the missing enzyme and the low cortisol level, the pituitary gland in the brain makes higher amounts of another hormone called **ACTH**, which tries to push the adrenal gland to make more cortisol. In this process, the adrenal glands make higher levels of androgens, a process not affected by the absence of this enzyme. The high androgen levels can cause changes in the genital area.

## WHAT ARE THE SYMPTOMS OF CAH?

Symptoms of CAH depend on your child's age:

### Newborns

Newborn babies may have a life-threatening condition called "**adrenal crisis**." Adrenal crisis can have the following symptoms:

- Vomiting
- Severe dehydration
- Low blood pressure
- Low blood sugar
- Shock
- Abnormal sodium and potassium levels

In females, the genitals might look more like male genitals.

In males, the genitals may look darker than expected.

In a less severe form (simple virilizing CAH), genital findings are present without salt (sodium and potassium) abnormalities.

### Older children

When CAH is diagnosed later in life, it is usually a milder form of the condition. This is called **Non-classic (late onset) CAH**. You might notice the following signs:

- Body odor, armpit hair or pubic hair before age 8 (in girls) or age 9 (in boys)
- Early growth spurt
- Early puberty

***Diagnosis and treatment info on the back! >>>***

## HOW DO DOCTORS DIAGNOSE CAH?

In most cases, the **newborn screening test** will help diagnose CAH at birth. If the care team thinks your child might have CAH on the newborn screening, they might do another blood test. The blood test checks for a hormone called 17-hydroxyprogesterone (17-OHP).

If your child has high levels of 17-OHP in the blood, the care team might have your child do an **ACTH stimulation test**. This test measures how well your child's adrenal glands make cortisol when your child is given cosyntropin, an artificial form of ACTH.

If CAH runs in your family, doctors can also diagnose CAH through **genetic testing**.

## HOW DO DOCTORS TREAT CAH?

Treatment for CAH includes:

- **Hormone replacement.** This can include replacing cortisol alone or with aldosterone. Hormone replacement medications are given by mouth. It is very important to take them every day or your child can get very sick. Cortisol is usually replaced using a medication called hydrocortisone 3 times per day. After your child is done growing, some doctors will switch to other medications such as prednisone or dexamethasone. Aldosterone is usually replaced using a medication called fludrocortisone.
- **Monitoring your child's bone age.** The care team will check your child's bone maturity through an X-ray of the left hand.
- **In babies, salt replacement.** Salt is replaced as a liquid or a pill. This (along with fludrocortisone) helps maintain normal salt levels in blood.

## WHERE CAN I LEARN MORE ABOUT CAH?

- Your child's care team at MGHfC
- Endocrine Society Hormone Health Network: [www.hormone.org](http://www.hormone.org)
- National Institutes for Health: [www.nichd.nih.gov](http://www.nichd.nih.gov)
- The Magic Foundation: [www.magicfoundation.org](http://www.magicfoundation.org)

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