CONGENITAL ADRENAL HYPERPLASIA (CAH)

THE FACTS YOU NEED TO KNOW

NADF
National Adrenal Diseases Foundation

What is Congenital Adrenal Hyperplasia?

Congenital adrenal hyperplasia, also called CAH, is a group of genetic disorders in which the two adrenal glands do not work properly. People inherit one gene that causes this disorder from each of their parents. This is known as a recessive genetic disorder. This means that carriers of the trait show no symptoms, but when one has a double dose of the trait, problems occur. The reason for these problems is that the adrenal glands, located on top of each kidney, cannot efficiently produce the hormones that are essential for body functions.

People with CAH lack one of the enzymes (proteins that cause chemical changes in the body), steroid 21-hydroxylase. This results in low production of the hormone (cortisol) that helps the body respond to stress, and in most cases of classic CAH they lack another hormone (aldosterone) needed to retain sodium. Individuals affected by classic CAH caused by 21-hydroxylase deficiency produce excess adrenal steroids that lead to the production of testosterone and related male-like hormones. CAH can be severe (classic) and diagnosed in the newborn, but it can also be mild (nonclassic) and not show up until later childhood, adolescence or adulthood.

Adrenal cortex hormones and their functions

Androgens (male sex hormones)
Control puberty, growth and male sexual characteristics.

Aldosterone (a mineralocorticoid)
Helps keep a proper balance of salt (sodium) and water in the body. Regulates blood volume and blood pressure.

Cortisol (a glucocorticoid)
Helps the body cope with stress, illness, and injury. Regulates blood glucose and blood pressure levels.

Estrogens (female sex hormones)
Control puberty, growth and female sexual characteristics.

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, all newborns undergo a blood test for the most common form of classic CAH. 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 before birth. Prenatal treatment of CAH is experimental. There is no cure for CAH, and giving steroids to the pregnant mother is merely an attempt to reduce fetal production of male sex hormones and thereby prevent the atypical genital appearance in affected baby girls with severe forms of CAH. Many experts are concerned about the potential for as yet unknown long-term side effects of using dexamethasone, a very strong steroid, during pregnancy—even if it is for a short time.

What are the different types of CAH 21-hydroxylase deficiency?

There are two types of CAH—classic, which can be life threatening, and nonclassic, 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. There are two forms:

- "salt-wasting" (meaning the body has trouble keeping the right amount of salt in the blood), in which the adrenal glands do not make enough cortisol and aldosterone.
- "non-salt wasting," in which the enzyme shortage is less severe and the adrenal glands make just enough aldosterone but not enough cortisol.

If not found and treated, classic CAH can cause shock, coma, and death.

SIGNS AND SYMPTOMS

In many cases, female infants are diagnosed at birth because they have atypical or 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. If a boy is not diagnosed in infancy, he may grow rapidly and show signs of early puberty.

Any infant, boy or girl, who is not diagnosed and treated appropriately at birth, may have vomiting, weight loss, dehydration, shock, and even death.

TREATMENT

The goals of treatment are to help maintain balanced hormone levels and promote normal growth in children. Concerns in adult patients revolve more around reproductive function. Patients with classic CAH should have a team of health care providers, including specialists in endocrinology, gynecology, urologic surgery, reproductive health, 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. Infants usually need sodium chloride (salt) supplements.

There is controversy about when and whether to perform surgery to alter the appearance of an enlarged clitoris or separate common vaginal and urethral openings in girls. There are inadequate long-term follow up studies comparing the effects of early surgery versus no surgery or delaying surgery until a child can consent for herself.

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, acne and/or 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. Nonclassic CAH is not associated with genital abnormalities at birth and is not detected in most newborn screening programs.

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.

Everyone with CAH should wear a medical alert bracelet stating they have the disease. An identification card outlining treatment is also suggested.

RESOURCE: **The CARES Foundation: www.caresfoundation.org**

Written by
Phyllis W. Speiser, MD
Medical Advisor, NADF
Chief, Division of Pediatric Endocrinology
Professor of Pediatrics
Cohen Children's Medical Center of New York
Hofstra Northwell School of Medicine

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The National Adrenal Diseases Foundation is a non-profit organization providing information, education and support to all persons affected by adrenal disease. For more information on joining NADF, or to find a support group in your area, contact:

The National Adrenal Diseases Foundation
P.O. Box 95149
Newton, MA 02495
(847) 726-9010

www.nadf.us
e-mail: NADFmail@nadf.us