

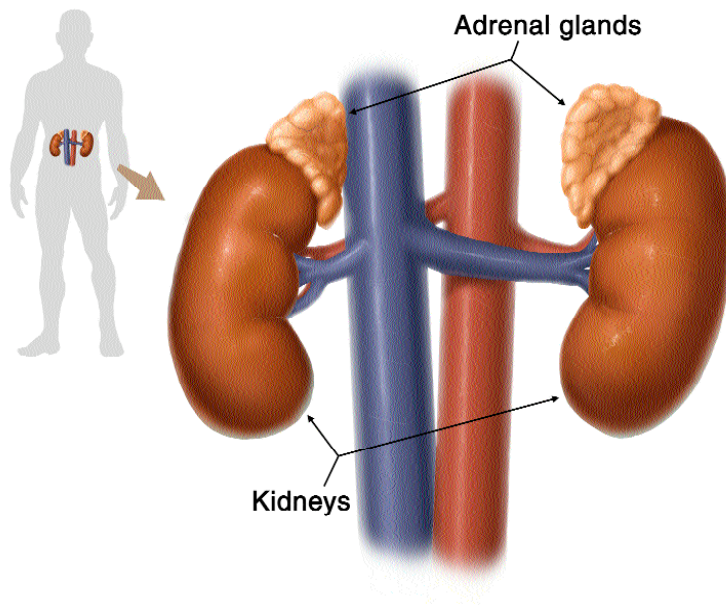


Facts about CAH Congenital Adrenal Hyperplasia

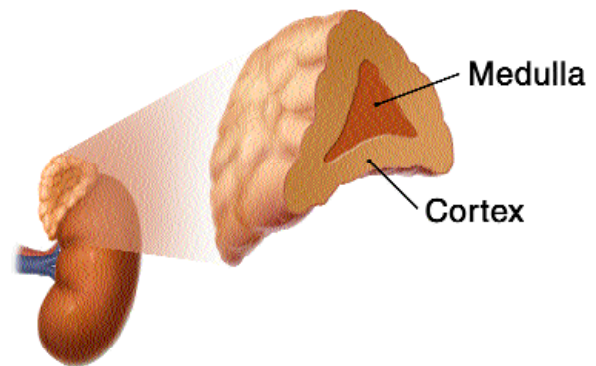
This information was prepared by your health care team to help you learn about congenital adrenal hyperplasia (CAH). CAH is a genetic disorder of the adrenal glands that affects the body's general health, growth, and development.

What are the adrenal glands?

The adrenal glands are a pair of walnut-sized organs above the kidneys. They make hormones, which act like chemical messengers to affect other organs in the body.



An organ at the base of the brain, called the pituitary gland, helps regulate the adrenal glands.



Each adrenal gland has two parts: the medulla (the inner part), and the cortex (the outer part). The medulla makes the hormone adrenaline. The cortex makes the hormones cortisol, aldosterone, and androgens.

CAH affects how the adrenal cortex works. In severe cases, the adrenal medulla may also not function normally.

What do adrenal hormones do?

Hormones made by the adrenal glands are important for the body's normal function. Cortisol affects energy levels, sugar levels, blood pressure, and the

body's response to illness or injury. Aldosterone helps maintain the proper salt level. Androgens are male-like hormones needed for normal growth and development in both boys and girls. Adrenalin affects blood sugar levels, blood pressure, and the body's response to physical stress.

What is CAH?

The adrenal glands help keep the body in balance by making the right amounts of cortisol, aldosterone, and androgens. But in CAH, production of cortisol is blocked. Some children with CAH also lack aldosterone. These imbalances cause the adrenal gland to make too much androgen.

Symptoms

Too little cortisol may cause tiredness and nausea. During illness or injury, low cortisol levels can lead to low blood pressure and even death.

Lack of aldosterone, which occurs in three out of four patients with classic CAH, upsets salt levels. This imbalance may cause dehydration (too little fluid within the body), and possibly death. Chronic salt imbalance may also cause abnormal growth.

Too much androgen causes abnormal physical development in children. Boys and girls with CAH may grow too fast, develop early pubic hair and acne, and stop growing too soon, causing

short stature. Girls exposed to high levels of androgens before birth may have abnormal external genitalia at birth. Although their internal female organs are normal, excess androgens may also affect puberty and cause irregular menstrual periods.

Too much cortisol replacement also causes abnormal development in children. Side effects include obesity and short stature. Also, too much hydrocortisone, the medicine given to replace cortisol in the body, can cause decreased bone density (osteoporosis) and high cholesterol levels.

Are there different types of CAH?

There are many types of CAH. The severe form is called classic CAH, while the mild form is called nonclassic CAH.

Classic CAH

The most common is 21-hydroxylase deficiency (95 percent of cases). A child with this type of CAH has adrenal glands that cannot make enough cortisol and may or may not make aldosterone. As a result, the glands over-work trying to make these hormones and end up making too much of what they can make: androgens.

The second most common form of CAH is 11-hydroxylase deficiency. A child with this type of CAH has adrenal glands that

make too much androgen and not enough cortisol. Children with this type of CAH may also have high blood pressure. These patients do not have aldosterone deficiency.

Rare other types of CAH include 3-beta-hydroxy-steroid dehydrogenase deficiency, lipoid CAH, and 17-hydroxylase deficiency.

Nonclassic (late-onset) CAH

This type of CAH is a mild form of CAH and is almost always due to 21-hydroxylase deficiency. Only a handful of people have been described as having nonclassic (mild) CAH due to other causes. People with nonclassic 21-hydroxylase deficiency make enough cortisol and aldosterone, but they make excess androgens. Symptoms come and go, beginning at any time but typically in late childhood or early adulthood. Boys often do not need treatment. Girls usually need treatment to suppress their excess androgens.

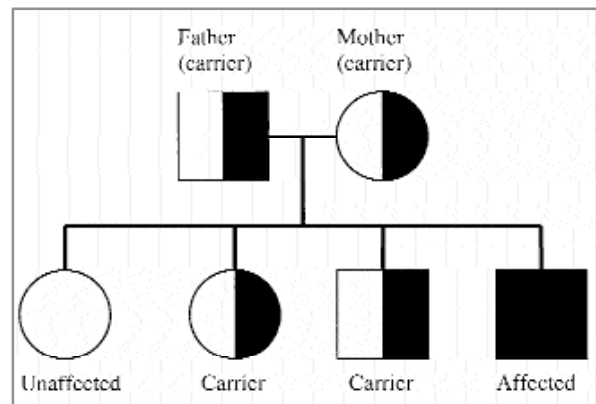
Nonclassic CAH is common. One in every 1,000 people has nonclassic 21-hydroxylase deficiency. Incidence is higher in certain ethnic groups including Ashkenazic Jews, Hispanics, Yugoslavs, and Italians.

How is CAH inherited?

An inherited disorder is one that can be passed from the parents to their children. CAH is a type of inherited disorder called “autosomal recessive.”

For a child to have CAH, each parent must either have CAH or carry a genetic mutation. This means that if two parents are CAH carriers (that is, they have the gene for CAH but not the disorder), their children have a 25 percent chance (1 in 4) of being born with CAH. Each sibling without CAH has two chances in three of being a carrier. Tests can be done to find out if someone is a carrier.

Classic CAH occurs in 1 in 15,000 births.



How is CAH treated?

The standard treatment for classic 21-hydroxylase deficiency is hydrocortisone which replaces cortisol, and fludrocortisone (Florinef) which replaces aldosterone. For 11-hydroxylase deficiency, the treatment is only hydrocortisone. Patients can be started on longer-acting forms of hydrocortisone (i.e. prednisone or dexamethasone) when they are done growing.

Because replacement medications cannot mimic the body's exact needs, patients, on average, are about 4 inches shorter than their peers.

Patients with the nonclassic form of CAH, need only hydrocortisone (or a longer-acting form of hydrocortisone). Some patients with nonclassic CAH are able to come off medication as adults, but patients with classic CAH need lifelong treatment.

What if a child with CAH has an illness, surgery, or a major injury?

During these times, a child with CAH needs closer medical attention and should be under a doctor's care. More cortisol is needed to meet the body's increased needs for this hormone. Higher doses of hydrocortisone are given by mouth or sometimes by intramuscular injection. Intravenous medication is needed before surgery.

Medical Alert Identification

In an emergency, it is important to alert medical personnel about the diagnosis of adrenal insufficiency, so wearing a medical alert identification bracelet or necklace is recommended. The information on the medic alert should include, "adrenal insufficiency, requires Cortef." It is also important for the adult or parent to learn how to administer an intramuscular injection of Cortef in case of emergency.

How long can people live with CAH?

People with CAH have a normal life expectancy.

Can a woman with CAH become pregnant and have a baby?

Increased androgens may cause irregular menstrual periods and make it harder for a woman with CAH to conceive a child. But if she takes her medications as directed, she can become pregnant and have a baby.

Do men with CAH have fertility problems?

Men who take medications as directed usually have normal fertility. Rarely, however, they may develop "adrenal rest tissue" in their testicles. This is when adrenal tissue grows in other parts of the body such as the testicles or scrotum. Having adrenal rest tissue may affect a man's ability to father a child. The tissue does not turn to cancer, but it can grow enough to cause discomfort or infertility. Large growths are rare, and surgery is usually not needed.

Do children with CAH outgrow it?

CAH cannot be outgrown. Classic CAH requires treatment for life. Some patients with nonclassic CAH may not require treatment as adults. Treatment is tailored for each patient and adjusted during childhood for growth.