

Congenital adrenal hyperplasia

Congenital adrenal hyperplasia (CAH) is a fairly uncommon genetic disorder, but it is well understood and treatment is readily available. CAH is also a congenital disorder (present at birth) involving the hormones of the adrenal glands. The word 'hyperplasia' means 'overgrown'. A child with CAH is born with overgrown adrenal glands.

What are the adrenal glands?

The adrenal glands are a pair of fleshy triangular-shaped organs, each about the size of a walnut, which lie above the kidneys, on the back wall of the abdomen. Although fairly small, the adrenal glands make some of the most important hormones that the body cannot do without, including cortisol. These hormones are available in tablet or injection form, so that the adrenal glands themselves can be done without if necessary.

In children with CAH, there is a problem with the complex chemical 'machinery' needed to make those essential hormones, and the glands make the wrong hormones.

Control of the adrenal glands

The adrenal glands are controlled by the pituitary gland, a pea-sized gland at the base of the brain. It is the 'master gland' that directs many of the other glands. When the adrenal glands are not producing enough of their main hormone (cortisol), the pituitary tells them to make more by sending an adrenal-stimulating hormone.

If the levels of adrenal-stimulating hormone remain high for too long, the adrenal glands grow larger. When too much cortisol reaches the pituitary, the pituitary switches off the adrenal glands, so they are allowed to rest until the hormone levels return to normal. Usually, the pituitary and the adrenal glands are perfectly balanced.

What are hormones?

Hormones are the chemical messengers in the body. They are produced in one place by an endocrine (or hormone-producing) gland, and act somewhere else in the body. Female sex hormone (oestrogen), for example, is made in the ovaries and acts elsewhere to cause breast enlargement, broadening of the hips and menstrual periods.

There are many different types of hormones and many endocrine glands, each making its own special hormones. In trying to understand CAH, the most important glands to consider are the adrenal glands and the pituitary gland.

Adrenal hormones

The adrenal glands produce cortisol, salt-retaining hormone and androgen. Not only are the adrenal glands each able to make all three hormones, but they make them from the same starting material – cholesterol. This can be made in the body, but is also found in the diet, in animal fats.

CAH results in three disturbances to these hormones:

- Lack of cortisol
- Lack of salt-retaining hormone
- Too much androgen.

Cortisol

Cortisol is needed to protect the body from the effects of illness or injury. If a person with poorly functioning adrenal glands develops an illness such as tonsillitis, or receives a significant injury such as a broken leg, they could go into a state of 'shock' (severe illness with dangerously low blood pressure) unless cortisol or an equivalent medication (such as cortisone) is given.

Salt-retaining hormone

This governs the amount of salt lost in urine via the kidneys. In the absence of this hormone, salt is lost uncontrollably, leading to dehydration and lack of salt.

Androgen (male sex hormone)

Both males and females have androgen. It is thought to aid growth in childhood and is responsible for women having pubic hair.

'Non-salt losers' and 'salt losers' and CAH

Children with CAH are classified as either 'non-salt losers' (20 per cent of CAH patients), or 'salt losers' (80 per cent).

Non-salt-losers

Children with non-salt-losing CAH are usually healthy, but may be born with genital abnormalities resulting from an excess of androgen. In girls, non-salt losing CAH is usually diagnosed at birth because the clitoris is large and the labia are partially fused. Sometimes, however, the changes may not be very obvious. In cases where the diagnosis is not made at birth, the growth of the clitoris continues and becomes much more obvious as time goes by.

In boys, non-salt-losing CAH produces nothing that can be detected at birth. Usually, the diagnosis is made when very prematurely the boy's penis enlarges, perhaps he grows some pubic hair, and he becomes unusually tall. These changes may not be noticed until two to three years of age. They all result from premature exposure to excessive levels of androgen. X-rays show advanced maturity of the bones. This is undesirable, and indicates that the child is closer to completion of growth than he should be. Some growth that should occur during childhood is lost. Because of this, adult height is usually somewhat reduced in people with this condition.

Salt-losers

In salt-losers, the deficiency in the adrenal glands is more severe than in the salt-losers. Their strong tendency to lose excessive amounts of salt in the urine, if uncontrolled, can cause acute dehydration, very low blood pressure, nausea and vomiting. The levels of salt (sodium and chloride) and sugar (glucose) in the blood fall, and the potassium level rises. This dangerous situation is referred to as an 'adrenal crisis'. Very urgent medical treatment is needed as a life-saving measure.

Some people with the salt-losing form of CAH receive very little warning of adrenal crises. Repetitive vomiting is often the first sign. Drowsiness is most likely to be due to an abnormally low blood sugar level, and if sugar is not given, may progress to loss of consciousness.

Non-salt-losers hardly ever experience adrenal crises because they make more cortisol and more of the salt-retaining hormone than the salt-losers.

What causes an adrenal crisis?

The body usually responds to sickness (fever, infection or other illness) and physical injury by releasing more cortisol. This helps to boost the blood sugar and blood pressure, aiding recovery. In CAH, the adrenal glands are unable to make more cortisol.

Therefore, the body is less protected and the effects of illness or injury may be more severe. Adrenal crises can be readily prevented.

Treatment of CAH

CAH is a lifelong disorder for which no cure is available. It is possible, however, to readily replace, in tablet form, the hormones that are missing – cortisol and the salt-retaining hormone. These medications will be needed every day for life, and need to be given even when the child is perfectly well. There is no need for a special diet with CAH. Girls with CAH require surgery to restore the genital appearance to normal. Boys do not.

Treatment for CAH

There are several medications available that are satisfactory substitutes for the natural hormone, cortisol. They are hydrocortisone, cortisone, prednisolone and dexamethasone. They differ in potency (strength) and duration of action. If a salt-retaining hormone is needed, fludrocortisone is used.

Dexamethasone is generally considered unsuitable for use in growing children because its high potency and very long action (24–48 hours) interferes with growth. Once adult height has been reached, however, dexamethasone provides excellent 24-hour control, with the added advantage that it needs to be taken only once a day.

Blood tests may be required every three months throughout childhood to help monitor replacement medication therapy. An injection of cortisone (hydrocortisone) may be required if a person with CAH becomes very sick or needs surgery.

When to give extra cortisone

Extra cortisone is not necessary with minor cuts and scratches, even if local anaesthetic (an injection near the cut to deaden sensation) is proposed. However, if the injury is more severe (such as a broken arm or leg), extra cortisone should be given.

Other times to use extra cortisone include:

- When your child feels ill, particularly if he or she has a high temperature
- When your child requires a general anaesthetic for any reason. It is absolutely essential that he or she is given an injection of cortisone beforehand. Failure to do this could cause death
- When your child is vomiting, or has diarrhoea. Not only does the child have to cope with the extra demands of being ill, but also the stomach may not absorb the medications needed to treat the CAH in the usual way. Your child may need to be given an injection of cortisone. Severe vomiting or diarrhoea in someone with CAH is often best treated in hospital.

Consult your doctor about how much extra cortisone to give your child. If you are in doubt when to give extra cortisone, it is always safer to give it, than not to give it.

Advice for people with CAH

Precautions for people with CAH include:

- Every person with CAH should wear an identification disc or bracelet carrying the words, 'Adrenal insufficiency: In emergency, give cortisone'.
- If you plan to travel, it is a good idea to ask your child's specialist for a letter outlining the important medical problems that your child could have, and how these should be dealt with in an emergency.

Genital changes in girls with CAH

In baby girls with CAH, the genitalia may look more masculine than they should. The clitoris is enlarged and may resemble a small penis. In addition, the cleft between the labia or lips may be partly closed over, hiding the entrance to the vagina. Often, only one opening can be seen. The urinary passage and vagina both open into this one entrance.

The internal organs, however, are quite normal. The vagina, uterus (womb) and ovaries are perfect, and girls with CAH are usually able to have children.

Sometimes it is difficult to be sure about a CAH baby's true sex at the time of birth. Tests to determine what internal organs are present may be needed, as well as hormone tests related to the adrenal glands. The results of these tests can usually be obtained within two to three days.

Surgical treatment for girls with CAH

Girls with CAH may need surgery to reduce the size of the clitoris to normal, separate the fused labia and enlarge the vaginal entrance. The technical name for this operation is 'clitoral reduction and vaginoplasty'. It is done either in one or two stages.

The clitoral reduction is done in the first few months of life. The vaginoplasty is sometimes done at the same time as the clitoral reduction, but may be left until adolescence, before the menstrual periods begin. Some stretching of the vagina may be necessary later to allow menstruation and comfortable sexual intercourse. The female hormones made by the ovaries at puberty soften the tissues and make the stretching easier.

Adult life for women with CAH

Women with CAH can have children of their own and there is little difficulty becoming pregnant provided that the hormone balance is kept under good control. If the hormones are not well balanced (perhaps due to forgetting to take medications), the menstrual periods may become irregular or even temporarily cease.

Because nearly all girls with CAH have had surgery around the vagina as children, there is scar tissue there. This may not stretch enough to allow vaginal delivery, and it is therefore sometimes necessary for a Caesarean operation to be performed to deliver the baby. Extra cortisone will be needed during labour and delivery.

How CAH is inherited

CAH is a genetic disorder resulting from one of the more common recessive gene defects. In fact, about one person in 50 in the general population has a CAH recessive gene hidden away among the many thousands of genes they possess, and yet only one person in 10,000 has CAH.

People with CAH have received a CAH gene from each of their parents. Because their cells only contain two abnormal recessive CAH genes and no normal ones, the cells in the adrenal glands are unable to make the enzyme in normal amounts, and this has a major effect on the way the adrenal glands work.

What are the odds?

If a couple already has one or more children with CAH, and neither parent has CAH, the chance with each subsequent pregnancy that the child will have CAH is one in four. If a person with CAH marries a person without CAH, the chance of their having a child with CAH is very low, about one in 100. If two people with CAH marry, all of their children would have CAH.

Where to get help

- Your doctor
- Endocrinologist
- Congenital Adrenal Hyperplasia Support Group of Australia Inc.

Things to remember

- CAH is a fairly uncommon genetic disorder, but one which is well understood and for which good treatment is readily available.
- CAH is a congenital disorder (present at birth) involving the hormones of the adrenal glands.
- The adrenal glands produce cortisol, salt-retaining hormone and androgen.

This page has been produced in consultation with, and approved by:

Congenital Adrenal Hyperplasia Support Group Australia Inc

Content on this website is provided for education and information purposes only. Information about a therapy, service, product or treatment does not imply endorsement and is not intended to replace advice from your doctor or other registered health professional. Content has been prepared for Victorian residents and wider Australian audiences, and was accurate at the time of publication. Readers should note that, over time, currency and completeness of the information may change. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions.

For the latest updates and more information, visit www.betterhealth.vic.gov.au

Copyright © 1999/2012 State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.