

Congenital adrenal hyperplasia

The term congenital adrenal hyperplasia (CAH) refers to any one of a group of inherited disorders in which adrenal gland hormones are either not being produced or are produced in incorrect amounts. Worldwide, approximately one in 14,000 babies are born each year with CAH. This condition is commonly diagnosed in infancy.

The role of the adrenal glands

The human body has two adrenal glands, one on top of each kidney. These glands form part of the endocrine (hormonal) system, which works in conjunction with the nervous system and the immune system to help the body cope with different events and stresses. Adrenaline is the most well-known hormone secreted by the adrenal glands. Other important hormones include:

Cortisol
Aldosterone
Sex hormone androgen.

All these hormones are synthesised from cholesterol, which is an oily substance manufactured by the body and found in animal, and some vegetable, products. Particular enzymes are needed to help convert cholesterol into the adrenal hormones.

The underlying problem in CAH is that not enough of these enzymes are produced. This is due to a genetic or inherited 'fault'. The symptoms of the disorder depend on the particular enzymes affected. There is no cure for CAH, but the disorder can be managed very well with medications.

The adrenal hormones

The hormones produced by the adrenal glands perform many vital bodily functions, including:

- **Cortisol** – helps the body cope with stress, maintains constant blood sugar levels, aids the immune system and contributes to blood pressure control.
- **Aldosterone** – maintains the correct amount of salt in the body, which influences blood volume and blood pressure.
- **Androgen** – one of the male sex hormones, responsible for male sexual characteristics.

A group of disorders

Congenital adrenal hyperplasia (CAH) is an inherited group of disorders that involves any one of the enzymes necessary to manufacture cortisol by the adrenal glands. The disorders are named after the specific hormone that is deficient:

- 21-hydroxylase deficiency (most common)
- 11-beta-hydroxylase
- 3-beta-hydroxysteroid dehydrogenase
- 17-alpha-hydroxylase (very rare).

21-hydroxylase deficiency is the most common

21-hydroxylase deficiency is the most common form of CAH. In more than 90 per cent of CAH cases, the steroid 21-hydroxylase is the defective enzyme. When this enzyme is defective, the production of cortisol is blocked.

Symptoms

When this enzyme is defective, it will cause:

- Production of cortisol to be blocked
- Overproduction of androgens
- An inability to produce the sodium (salt) regulating hormone, aldosterone (in approximately 75 per cent of 'classical' cases).

Classical CAH

The most severe type of CAH, classical CAH, is often described as being either 'salt-losing' or 'non-salt-losing'. Both these forms of CAH are characterised by insufficient cortisol production and androgen excess. In addition, salt losers produce no aldosterone.

'Salt losers'

Around eight out of 10 children with classical CAH are 'salt losers'. Insufficient amounts of the hormone aldosterone mean that the child's kidneys excrete too much salt. A molecule of salt attracts many molecules of water. If the kidneys excrete too much salt, a considerable amount of body fluid is also lost. This reduces blood volume and cause a drop in blood pressure. Other symptoms include:

- Dehydration
- Vomiting
- Nausea.
- 'Classical salt-losing CAH' is sometimes referred to as 'salt-wasting CAH'.

Adrenal crisis is life threatening

A side effect of plummeting salt in the blood is a rise in potassium levels. This condition is known as 'adrenal crisis' and can be life threatening without prompt emergency treatment.

'Non salt losers'

Around two out of 10 children with classical CAH are 'non salt losers'. These children have sufficient aldosterone, so there is no associated salt loss. However, their adrenal glands do not produce enough cortisol and produce too much of the male sex hormone androgen. This has a masculinising effect:

- Baby girls are often diagnosed at birth, because the action of the male hormone causes female genital (clitoral) enlargement and partial fusion of the labia, or lips of the vulva.
- Boys are generally diagnosed later, when their penis and pubic hair develops at an abnormally early age, such as two or three years.

The bones of both girls and boys tend to fuse early, which results in reduced adult height if they do not receive the correct treatment. Classical 'non-salt-losing CAH' is sometimes called simple 'virilising CAH'.

Non-classical 'late onset'

Non-classical CAH is a milder and more common type of the condition. Although non-classical CAH also results from an inherited gene mutation, the enzyme deficiency is less severe than in the classical form. The signs and symptoms of late onset (non-classical) CAH may start at any age, through to and including adulthood. Some of the symptoms may be:

- Early growth of pubic hair
- Body odour in childhood
- Facial hair
- Persistent acne
- Irregular menstrual cycle
- Infertility (male and female).

Depending on the hormone levels and the symptoms, treatment may include corticosteroids (cortisone medication) to correct the androgen excess. Other treatments may also be used.

Tips for parents and caregivers

Parents and caregivers of people with CAH need to be able to recognise situations that may lead to an adrenal crisis. They should seek medical help immediately and can then, in consultation with their doctor, give appropriate doses of the required medication to prevent a crisis from occurring.

Both 'salt losers' and 'non salt losers' are at risk of severe shock and circulatory collapse if they experience major body stress or get very sick. Seek specialist medical advice if any of these occur:

- Broken bones
- Surgery
- Diarrhoea and vomiting

- Tonsillitis.

Reconstructive surgery for girls with CAH

Female infants born with either salt-losing or non-salt-losing CAH commonly have ambiguous (not clearly female or male) genitals due to an excess of the male sex hormone androgen. A baby girl with CAH is usually born with an enlarged clitoris and partly fused labia. Surgery is usually performed early in life. The dual procedures include:

- **Separating the labia** – the resulting scar tissue may constrict and narrow the opening of the vagina.
- **Trimming the clitoris** – great care is taken not to disturb nerve pathways or blood vessels, to ensure the best possible functioning of the clitoris.

Long term management for women with CAH

The surgery and resulting scars tend to narrow the vaginal opening. Hormones produced at puberty will help the skin to become supple. The regular use of tampons instead of sanitary pads during the menstrual period can further help to dilate the vagina.

Young women with CAH may need to be examined at some stage by a gynaecologist to make sure the vagina is structurally capable of sexual intercourse. A procedure can be performed where a narrowed vagina can be gently stretched with dilators, or a subsequent operation can be performed.

Medications

There is no cure for CAH, but the condition can be well managed with replacement cortisol and a synthetic salt-retaining hormone treatment. Synthesised versions of the missing adrenal hormones are taken every day in tablet form.

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.

Where to get help

- Your doctor
- Specialist endocrinologist
- Congenital Adrenal Hyperplasia Support Group Australia Inc. Tel. (03) 9513 9255

Things to remember

- Congenital adrenal hyperplasia (CAH) is an inherited group of disorders characterised by incorrect amounts of adrenal gland hormones.
- CAH is a genetic disorder, caused by an insufficiency of the particular enzymes that help convert cholesterol into the adrenal gland hormones.
- There is no cure, but the condition can be managed with medications. Most people with CAH enjoy good health and a normal lifestyle.

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

Congenital Adrenal Hyperplasia Support Group Australia Inc

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