Congenital Adrenal Hyperplasia

Series N. 6

Patient's Guide

Average Readability Leaflet

Congenital Adrenal Hyperplasia - Series 6 (Revised August 2006)

This leaflet was produced by Fernando Vera MSc and Prof Gary Butler at the Institute of Health Sciences, University of Reading, Reading, UK (August, 2006). Some portions of the text were extracted or modified from the Growth and Growth Disorders Booklet Series (Third edition, 2000)* and may be used in conjunction with these as they provide a choice of leaflets providing the same information, but for people of different ages and reading abilities. The numbering sequence in each series is the same for easy cross-reference. The original leaflet series can be also obtained from the links given at the end.

All illustrations were created and produced by Fernando Vera MSc.

This leaflet is part of the Hormone Disorders Leaflet Series. The following are also available:

Series N 3. Puberty and the Growth Hormone Deficient Child.

Series N 4. Precocious Puberty

Series N 5. Emergency Information for Children with Cortisol and GH Deficiencies and those Experiencing Recurrent Hypoglycaemia.

Series N 6. Congenital Adrenal Hyperplasia

Series N 7. Growth Hormone Deficiency in Young Adults.

Series N 10. Constitutional delay of growth and puberty

Series N 11. Multiple Pituitary Hormone Deficiency

Series N 12. Diabetes Insipidus

Series N 13. Craniopharyngioma

Series N 14. Intrauterine Growth Retardation or Small Gestational Age

Series N 15.a. Hyperthyroidism

Series N 15.b. Hypothyroidism

Series N. 16. Type 2 Diabetes and Obesity

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*Written by Dr Richard Stanhope (Gt. Ormond Street/Middlesex Hospital, London) and Mrs Vreli Fry (Child Growth Foundation

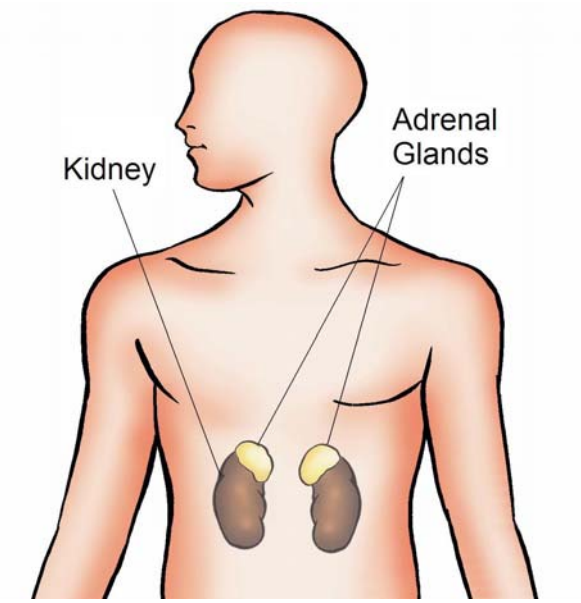
Introduction

The aim of this leaflet is to describe a condition known as Congenital Adrenal Hyperplasia. It will discuss information on how it's diagnosed, treated and some of the problems it may cause.

Hopefully, this leaflet will help you to understand this condition and give you a basis for discussions with your GP or specialist team.

What are the adrenal glands?

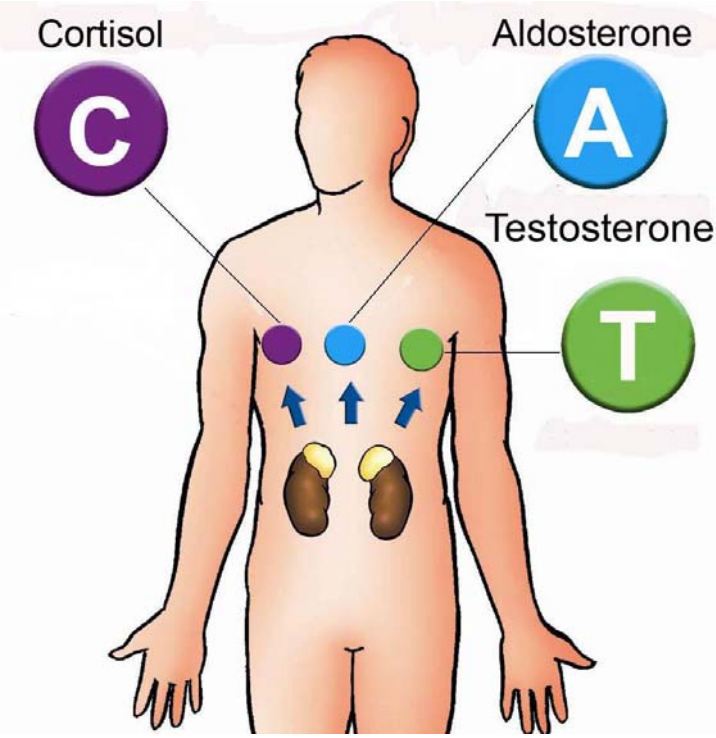
The adrenal glands are a pair of triangular shaped organs, which lie above the kidneys.



The adrenal glands work in conjunction with the brain to produce hormones that are essential for normal health.

These hormones are three main types:

- **Cortisol:** Helps the body to combat illness and stress. It's also in charge of maintaining the proper level of sugar in the blood.
- **Aldosterone:** Controls the salt and water balance in the body.
- **Testosterone:** This is a male hormone (androgen) responsible for the development of male characteristics



What is Congenital Adrenal Hyperplasia?

Congenital Adrenal Hyperplasia (CAH) is a condition in which the adrenal glands fail to make sufficient cortisol and/or aldosterone, and produce excess androgen. This condition is inherited (passed in families) and it's present before birth.

How does the common form of CAH affect children*?*

The way in which the common form of CAH is expressed depends upon whether the child is a boy or a girl, and whether the deficiency is severe or mild.

- **In boys with severe deficiency:** The baby will look normal at birth. The first signs of CAH are usually poor feeding, weight loss and vomiting during the first and second week. This is due to salt and water loss from aldosterone deficiency.

- **In girls with severe deficiency:** The baby will have been exposed to excessive amounts of male hormones while inside the womb. This causes the genitalia to appear masculinised (male-like). However, the baby is definitely a girl with a normal uterus, vagina, and ovaries internally.

  If the diagnosis of CAH is delayed, these baby girls can have the same salt-losing problems as boys. Children (male and female) with this type of **classical** CAH are often termed **salt-losers.**

- **Boys with mild deficiency:** They will present later (between two and four years of age) with tall stature, enlargement of the penis, and perhaps some pubic hair. These effects are due to excessive amounts of male hormones.

- **Girls with mild deficiency:** They will also show the effects of too much male hormone. They will be tall with pubic hair and some enlargement of the clitoris. These more mildly affected boys and girls are called **non-classical**.

Although these boys and girls are tall for their age, they will become short adults if left untreated. This is because the male hormones have the effect of making the bones mature rapidly, so that growth finishes earlier than normal.

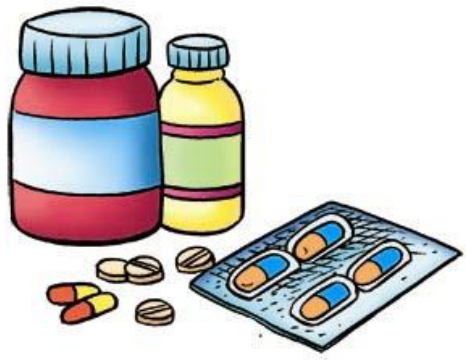
Finally, there is a very mild form of **non-classical** CAH, which can cause excess body hair and irregular periods in young women.

What treatment is required in CAH?

Treatment for children with CAH can be divided up into **medical** treatment and **surgical** treatment.

Medical treatment

Medical treatment for CAH consists of giving cortisol to correct any deficiency. This can be given as **hydrocortisone** in the form of tablets. For those children, who are also aldosterone deficient, a substitute hormone will be given. This is usually **fludrocortisone** tablets.



Extra salt is sometimes given to babies with severe CAH **salt-losers).**

The frequency of giving medicine in CAH varies from child to child. Under certain circumstances such as illness or severe stress, hydrocortisone needs to be given by injection.

Surgical treatment

Surgical treatment is only required in some girls. This is usually carried out when the child is about one year of age. The nature of surgery required depends on the degree of masculinisation. The surgeon will

need to reduce the size of the clitoris, whilst preserving the delicate supply of nerves and blood vessels. Also, the surgeon may try to open the entrance to the vagina.
Once puberty has started, these girls should be referred to the original surgeon, or to a specialist gynaecologist. They will determine if any further surgery is going to be needed in adolescence.

Is puberty normal in CAH?

In children with well-managed CAH, puberty can be expected to start at the usual time and proceed normally. However, girls may to start their periods late with a possibility of developing polycystic ovaries. To achieve normal menstruation, the hormone doses may require some fine-tuning.

Some children with other rare forms of CAH do not have a normal pubertal development. In this case, sex hormones will be given at the appropriate time. Since the uterus and ovaries are normal in girls with CAH, there is no reason why they should not have children of their own and a number of women have had normal pregnancies.

What follow-up tests are needed?

Tests must be carried out to measure hormone treatment doses. Blood tests and blood pressure measurement are used to estimate the doses for fludrocortisone. To estimate hydrocortisone doses, the body size, growth rate with or without blood/urine tests are used.

Additionally, a **bone age test** may be done. This consists of taking an x-ray of the hand and wrist to measure bone development. If bone development is advancing too fast this may require a higher dose of hydrocortisone.

How does the management of CAH change in an emergency?

Hydrocortisone is a very important hormone to combat illness and stress. Under these conditions, individuals with CAH will require an increase in their medication. Your doctor or nurse will advise you about what to do if your child is ill. In addition, please refer to leaflet Series No. 5 "Emergency Information for Children with Cortisol and GH deficiency".

What are other sources of useful of information?

The goal of this leaflet was to provide a basic overview of CAH. Further information can be found in the following sources:

- **European Society for Paediatric Endocrinology**
  ESPE Secretariat, BioScientifica
  Euro House 22 Apex Court Woodlands, Bristol BS32 4JT - UK
  Telephone No: + 44 (0) 01454 642208
  Internet: http://www.eurospe.org/
- **British Society for Paediatric Endocrinology and Diabetes**
  BSPED Secretariat, BioScientifica
  Euro House 22 Apex Court Woodlands, Bristol BS32 4JT - UK
  Telephone No: + 44 (0) 01454 642208
  Internet: http://www.bsped.org.uk/
- **Child Growth Foundation**
  2 Mayfield Avenue, Chiswick London W4 1PW UK.
  Telephone +44 (0) 20 8995 0257
  Internet: http://www.childgrowthfoundation.org/

You can also consult your doctor or nurse for additional information in your local area.