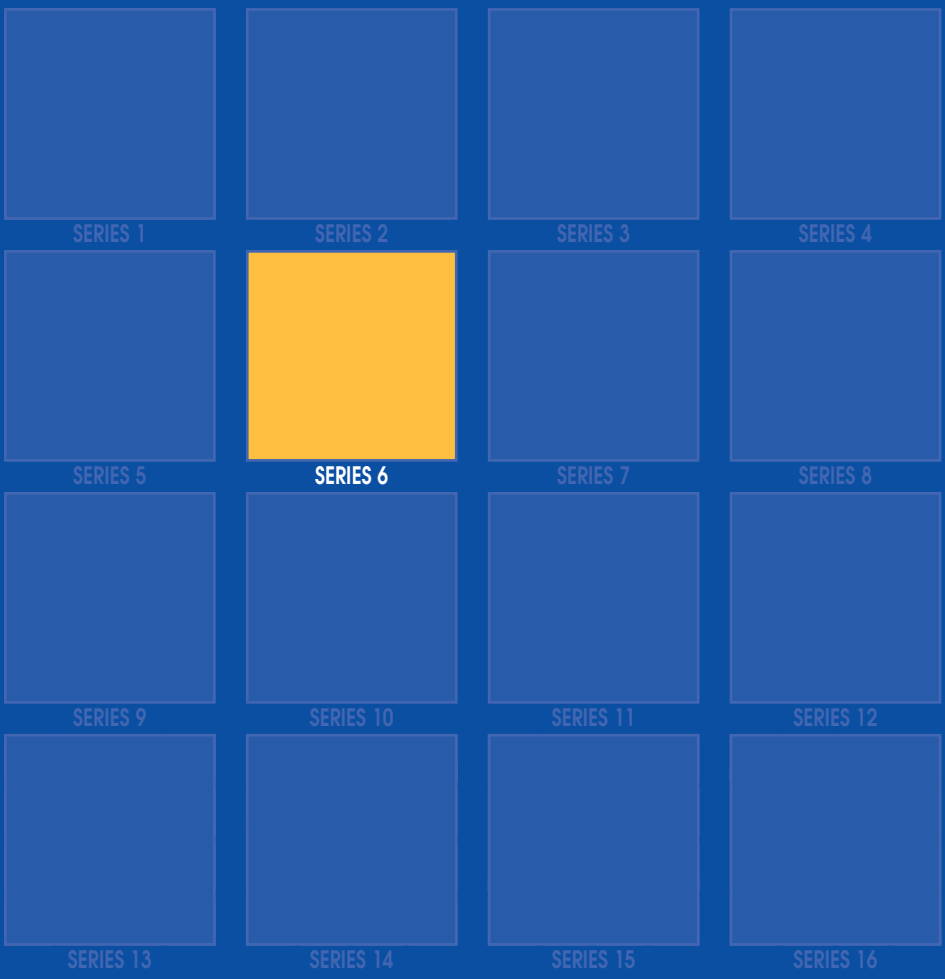
Congenital Adrenal Hyperplasia

A Guide for Parents and Patients



CHILD GROWTH FOUNDATION

BSPED

This booklet is intended to provide help when dealing with problems or difficulties associated with your child's condition and to provide information which will enable you to understand your child's treatment better and give you a basis for discussions with your child's specialist when necessary.

If you require further general information about *Congenital Adrenal Hyperplasia* you can contact the Child Growth Foundation.

THE CHILD GROWTH FOUNDATION
21 Malvern Drive
Walmley
Sutton Coldfield, B76 1PZ

Telephone: +44 (0)20 8995 0257
Email: info@childgrowthfoundation.org
www.childgrowthfoundation.org

CONGENITAL ADRENAL HYPERPLASIA – Series No 6 (February 1994), Revised January 1996

Prepared by Dr M D C Donaldson (Royal Hospital for Sick Children, Glasgow) and Dr D B Grant (Great Ormond Street, London). Edited by Mrs Vreli Fry (Child Growth Foundation) and Dr Richard Stanhope (Great Ormond Street Hospital for Sick Children and The Middlesex Hospital, London).

The British Society for Paediatric Endocrinology and Diabetes (BSPED) is an association of specialists who deal with hormone disorders in children.

CGF INFORMATION LEAFLETS

The following titles are also available:

Series No 1	Growth and Growth Disorders
Series No 2	Growth Hormone Deficiency
Series No 3	Puberty and the Growth Hormone Deficient Child
Series No 4	Premature Sexual Maturation
Series No 5	Emergency Information Pack for Children with Cortisol and GH Deficiencies and those Experiencing Recurrent Hypoglycaemia
Series No 6	Congenital Adrenal Hyperplasia
Series No 7	Growth Hormone Deficiency in Adults
Series No 8	Turner Syndrome
Series No 9	The Turner Woman
Series No 10	Constitutional Delay of Growth and Puberty
Series No 11	Multiple Pituitary Hormone Deficiency (MPHD)
Series No 12	Diabetes Insipidus
Series No 13	Craniopharyngioma
Series No 14	Intrauterine Growth Retardation (IUGR)
Series No 15	Thyroid Disorders

CONGENITAL ADRENAL HYPERPLASIA

Contents

INTRODUCTION

All parents expect their children to be healthy and most are deeply shocked when they learn that their child has a medical disorder which will require lifelong treatment. Often they have difficulty in remembering some of the things that they were told while their child was in hospital and it may be many months before they feel that they can discuss their worries with their doctor, or even with other members of the family.

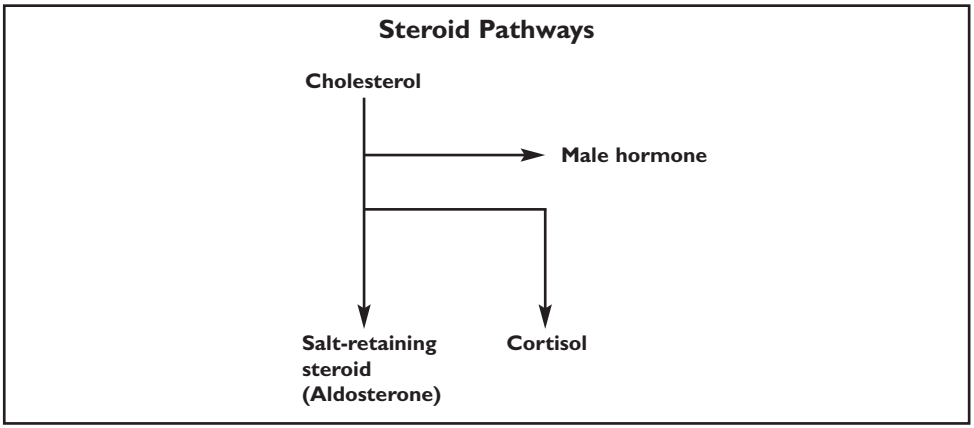
These notes may help answer some questions about Congenital Adrenal Hyperplasia. As you read them, please remember that every patient is slightly different and treatment may not be exactly the same in each case.

WHAT DOES CONGENITAL ADRENAL HYPERPLASIA MEAN?

The term Congenital Adrenal Hyperplasia, which is often abbreviated to CAH, describes what the adrenal gland looks like in this disorder and can be translated as thickening or overgrowth (hyperplasia) of the adrenal glands from before birth (congenital). The adrenal glands are a pair of flat organs, not much bigger than broad beans, which lie close to the kidneys deep in the abdomen. The adrenal gland is composed of an inner half called the **adrenal medulla** which produces **adrenaline** and works quite normally in CAH, and an outer half called the **adrenal cortex** which does not function correctly in patients with this condition.

WHAT DOES THE ADRENAL CORTEX NORMALLY DO?

The adrenal cortex produces a number of hormones called steroids which are secreted into the bloodstream and which are essential for normal health. These steroids are made from cholesterol and there are three main types – **cortisol**, **aldosterone** (salt-retaining steroid), and **androgen** (male hormone).



Cortisol, which is also known as hydrocortisone, keeps many of the body's systems, including the circulation and control of blood sugar level, in good working order. Cortisol is particularly important in helping the body combat stress (such as infection or injury) and in raising a low blood sugar level (hypoglycaemia).

The chief salt-retaining steroid is called **aldosterone** and this hormone helps to regulate the salt levels in the body. The amount of salt we take in each day varies with what we eat, and it is up to the kidneys to prevent accumulation of salt in the body (which would cause serious problems such as high blood pressure or fluid retention), while at the same time preventing excessive salt loss (which would cause vomiting and dehydration). Aldosterone makes the kidney conserve salt so that when we are taking very little salt in our diet aldosterone production is switched on. If we are taking a lot of salt, the adrenal cortex reduces the amount of aldosterone produced, allowing salt to be excreted in the urine.

Androgen (or male hormone) is produced by the adrenal cortex in both sexes, and also by the testes in the male. In both sexes, adrenal androgens contribute to the formation of pubic hair during normal puberty.

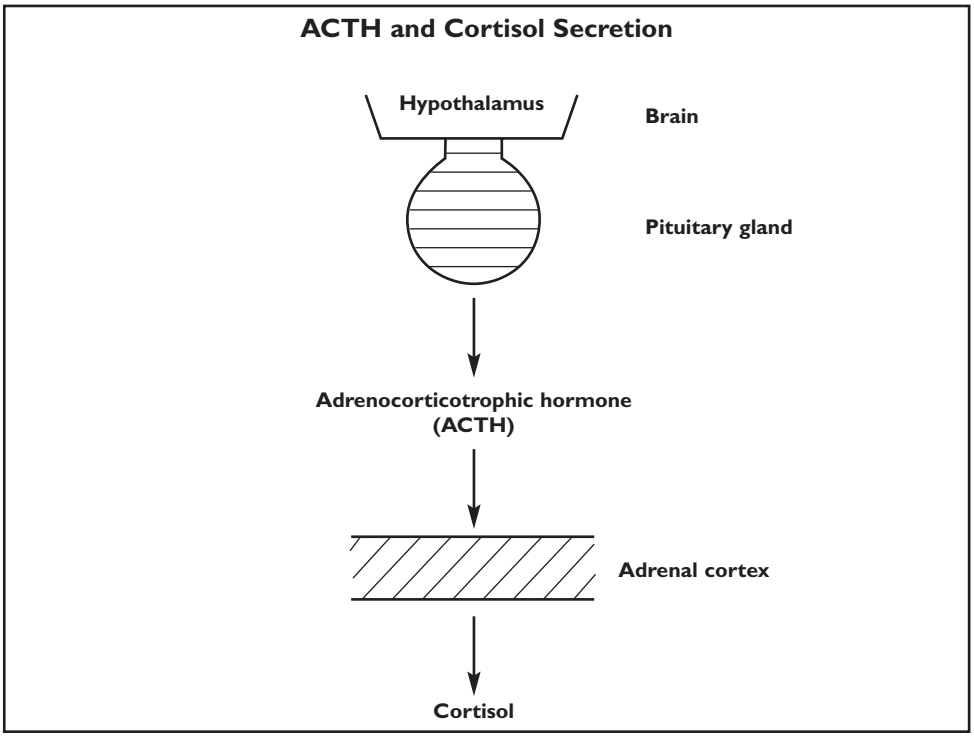
HOW DOES THE ADRENAL CORTEX KNOW HOW MUCH CORTISOL TO PRODUCE?

The amount of cortisol produced by the adrenal cortex is controlled by another organ, the pituitary gland, which lies in the base of the skull and is connected to a part of the brain called the hypothalamus. When the blood does not contain enough cortisol for the body's needs, the brain senses this and the hypothalamus makes the pituitary gland release another hormone called corticotrophin or adrenocorticotrophic hormone (ACTH).

The ACTH enters the blood stream, reaches the adrenal cortex and stimulates the production of more cortisol.When the cortisol concentration in the blood has been corrected, the brain recognises this and the hypothalamus stops stimulating the pituitary gland to release ACTH into the circulation. If the amount of cortisol in the blood is increased by giving a patient **cortisol treatment**, the pituitary gland is not stimulated to release ACTH and the adrenal cortex is not stimulated to release cortisol. This last point is very important in the treatment of CAH.

WHAT IS CAH?

The process of making cortisol from cholesterol in the adrenal cortex requires a complicated series of changes, each of which is brought about by a protein called an 'enzyme'. CAH occurs when any one of five enzymes is not working properly so that cortisol production is impaired. The pituitary gland senses the low levels of cortisol and



tries to compensate for this by secreting more ACTH. This in turn causes the adrenal cortex to increase in thickness and become 'hyperplastic'. In the common form of CAH, caused by a deficiency of the enzyme 21-hydroxylase, the production of both cortisol and aldosterone is impaired while the male hormone continues to be produced. This means that the continuous high level of ACTH secretion stimulates the adrenal cortex to make excessive amounts of male hormone. Thus, depending on the severity of the enzyme defect, CAH due to 21-hydroxylase deficiency results in **deficiency** of cortisol and aldosterone, with **excess** of male hormones.

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 enzyme deficiency is severe or mild.

In boys with a severe enzyme deficiency the baby will look normal at birth and the first signs of CAH are usually poor feeding, weight loss and vomiting occurring between the first and second week. This is due to salt and water loss in the urine from aldosterone deficiency which requires urgent treatment.

In girls with a severe enzyme deficiency the baby will have been exposed to excessive amounts of male hormones while inside the womb, causing the genitalia to appear masculinised (male-like). Thus, the outer lips of the entrance to the vagina (the labia) tend to come together to look like a scrotum, while the clitoris enlarges so that it can be mistaken for a male penis. Sometimes the masculinisation is sufficiently severe to make it unclear at first glance whether the baby is a girl or a boy, and this is a very distressing situation for the parents. However, the baby is definitely a girl who has female chromosomes (XX), and a normal uterus, vagina, and ovaries internally, but she will need surgery to correct the appearance of her external genitalia. If the diagnosis of CAH is delayed, these baby girls are prone to the same salt-losing problems as boys. Children (male and female) with this type of CAH are often termed '**salt-losers**' or '**salt-wasters**'.

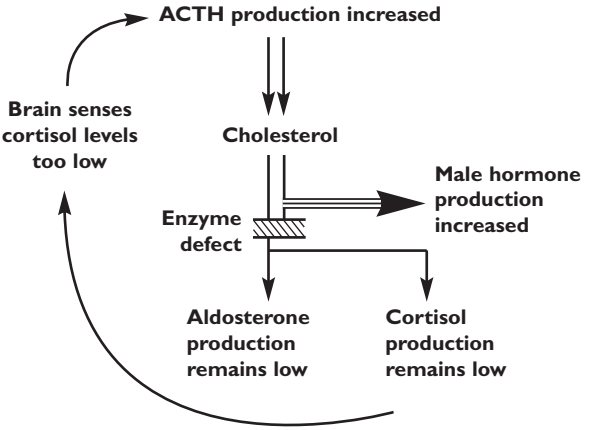
Boys with a mild enzyme deficiency will present later (often between two and four years of age) with tall stature, enlargement of the penis, and perhaps some pubic hair. These effects are due to excessive amounts of male hormones.

Girls with a mild enzyme deficiency will also show the effects of too much male hormone. They will be tall with pubic hair and some enlargement of the clitoris.

Although these boys and girls are tall for their age they will, if untreated, be relatively short as adults since the male hormones have the effect of making the bones mature rapidly, so that growth finishes earlier than normal.

These more mildly affected children do not have the same problem with salt loss as children with the more severe variety and are termed '***non-salt-losers***', although careful chemical testing will usually show a mild degree of salt depletion.

The common variety of CAH (21-hydroxylase deficiency)



Finally, there is a very mild form of CAH which is termed '***non-classical***' or '***cryptogenetic***'. This form is so mild that it does not usually present in childhood at all but can cause excess body hair and menstrual irregularity in young women. Often in these cases the ovaries will be enlarged with cysts (polycystic ovaries) and this is also commonly seen in girls with the salt-losing and non-salt losing variety of the condition.

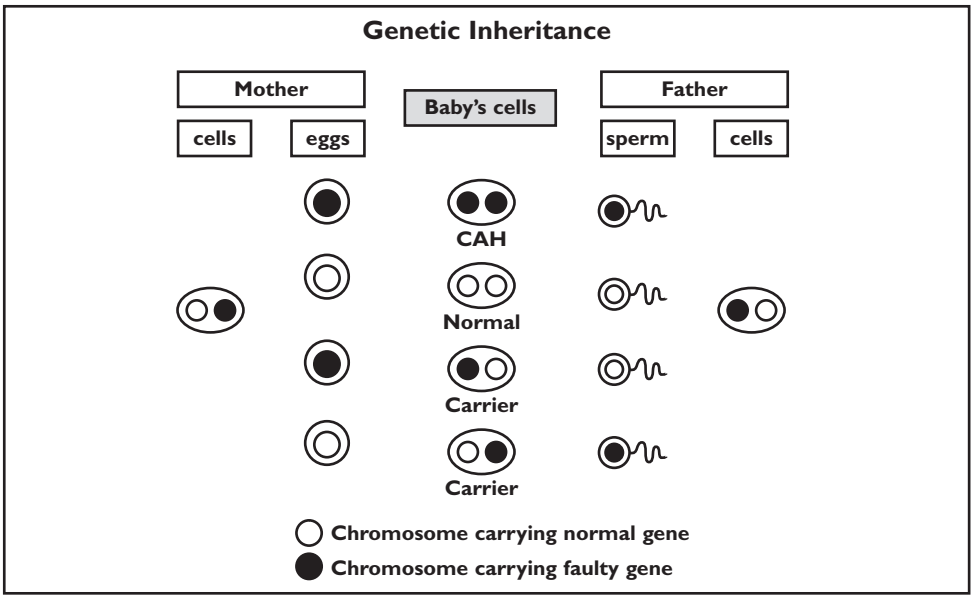
HOW DOES CAH HAPPEN?

CAH is an inherited disorder, and to understand how it is passed on you need to know a little bit about chromosomes and genes.

Babies are made when a sperm from the father fertilises an egg from the mother, forming a single cell. This cell then multiplies and the millions of cells that result form into various tissues (brain, muscle, liver, skin, etc).

The information required for an individual cell to produce a complete person is contained in tiny units called **genes**. These genes are stored on thread-like structures known as **chromosomes**. Each cell contains 23 pairs of chromosomes, one of each pair coming from the father, and one from the mother.

In the common form of CAH, one of the genes on chromosome 6 is faulty so that the enzyme 21 hydroxylase cannot be made properly. Parents whose child has CAH will have a faulty copy of the gene on one chromosome 6 and a normal copy of the gene on their other chromosome 6. Such parents are termed 'healthy carriers'. If a sperm carrying the



abnormal gene meets an egg which also carries an abnormal gene, then the baby will have inherited two faulty copies and, as a result, develops CAH.

If an abnormal sperm meets an normal egg (or vice versa), the baby will be a healthy carrier like the parents. A baby produced by the fusion of a normal sperm and an normal egg will, of course, not even be a carrier.

If you look at the figure on the opposite page again, you will see that out of four children born to parents who carry the CAH gene, the **chances** are that **one** will be affected by CAH, two will be carriers, and one will be neither affected nor a carrier.

This type of inheritance, where there is a 1 in 4 chance of having an affected child, is known as 'recessive'. It is important to stress that, if the parents are carriers, the risk of having an affected child is 1 in 4 for **each** pregnancy, so that having one child does not mean that the next three children will be unaffected.

ARE THE GRANDCHILDREN LIKELY TO HAVE CAH?

When a person with CAH grows up and has children, the chances are that their partner will not be a carrier for the condition. Although children born to that couple will inherit one faulty copy of the gene from the CAH parent, this will be balanced by the normal copy from the other parent.

Like their grandparents, therefore, these children will be healthy carriers. The risk of someone with CAH having a partner who is a carrier for the condition is about 1 in 50, but this risk will be higher if the couple are close blood relations such as cousins.

If someone with CAH does marry a carrier, then their risk of having an affected child is increased to 1 in 2.

IS PUBERTY NORMAL IN CAH, AND CAN GIRLS WITH CAH HAVE CHILDREN OF THEIR OWN?

In children with well managed CAH, puberty can be expected to start at the usual time and proceed normally. However, there is a tendency for girls to start their periods late and ultrasound examination will often show polycystic ovaries. To achieve normal menstruation the steroid doses may require some fine tuning, and this is one of the reasons why it is important for anyone with CAH to be followed up in the adult clinic once they have left paediatric care.

Some children who have other rare forms of CAH (not 21-hydroxylase deficiency) do not have a normal pubertal development and appropriate sex hormones will need to be administered at the appropriate time. If this applies to your child, your specialist will discuss this with you in detail.

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. However, girls with a strong salt-losing tendency may experience some difficulty conceiving, such that careful supervision of their treatment will be required when they want to start a family.

CAN CAH BE DETECTED IN THE UNBORN BABY, AND CAN ANY TREATMENT BE OFFERED?

If a family have had one child with CAH it is now possible, by taking blood from the child and parents and by testing tissue or amniotic fluid from the unborn baby to find out whether the baby is a boy or a girl and whether or not the baby has CAH. Also by taking blood from brothers and sisters, it is possible to find out whether or not they are carriers, although this information is not usually needed during childhood.

If the foetus is a girl with CAH, it is desirable to prevent the baby's external genitalia from becoming masculinised from excessive male hormone production. This can now be done by giving the mother a steroid called dexamethasone which is taken in tablet form and which passes from the mother's blood stream, across the placenta, into the baby's blood stream, stopping the baby's pituitary gland from producing too much ACTH. This treatment only works well if it is started at a very early stage, preferably as soon as the woman is aware of her pregnancy.

In families wishing to have this treatment we advise that the mother starts dexamethasone as soon as possible after she knows she is pregnant. Between the 10th and 14th week of pregnancy a piece of tissue is taken through the neck of the womb (chorionic villous biopsy) so that the sex of the baby can be determined, as well as diagnosing the presence or absence of CAH. If the pregnancy is too far advanced to take a biopsy, then amniotic fluid is sampled instead.

If the baby is unaffected, the dexamethasone treatment can be stopped. There will have been no ill effects for the baby from this short period of treatment. If the baby is an affected boy, the treatment can also stop since excessive male hormone production in a male baby is not a problem. If, however, the baby is an affected female, then the mother carries on taking the dexamethasone throughout pregnancy as this has been shown to diminish the amount of masculinisation, so reducing the extent of corrective surgery that may be required later on. So far as we know, there are no ill effects from this treatment on the baby, although the mother may experience more in the way of weight gain and stretch marks than usual.

WHAT TREATMENT IS REQUIRED IN CAH?

Treatment for children with CAH can be divided up into **medical** treatment with steroids and salt, and **surgical** treatment which will be required by most girls.

Medical treatment

Medical treatment consists of giving ***cortisol*** or an equivalent steroid to correct any deficiency and to stop the pituitary gland from producing too much ACTH. Under stress for example when the child is ill, the hydrocortisone dosage needs to be increased since the normal adrenal cortex responds by making more cortisol. This is particularly important to prevent the blood sugar from falling too low, a condition known as hypoglycaemia.

Cortisol can be given either in the form of cortisol itself (hydrocortisone), or as cortisone acetate, prednisone, or dexamethasone. These four steroids vary in strength but all are suitable for the treatment of CAH. They are widely available and relatively cheap, and all are available as tablets. Prednisone and dexamethasone are more potent than the other two steroids and tend to be used in older children. For the sake of brevity, we will refer to ***hydrocortisone*** from now on when discussing cortisol replacement.

In children who show signs of salt loss due to insufficient production of aldosterone, treatment with hydrocortisone alone is not enough. These children require a substitute hormone, similar to aldosterone, and this is usually given as ***fludrocortisone*** tablets.

In the first year of life, children with a salt-losing tendency will require an extra supplement of salt, usually added to feeds. After the first year, however, the combination of fludrocortisone and normal dietary salt is usually enough to keep the salt balance normal.

The frequency of giving medicine in CAH varies from centre to centre and from child to child. Mostly, the hydrocortisone or equivalent steroid is given twice or three times a day (although dexamethasone can be given just once a day), while fludrocortisone is usually given once a day, and salt several times a day.The frequency needs to be tailored to the individual child.

Under certain circumstances, e.g. if the child has been vomiting, is very ill or unconscious, hydrocortisone needs to be given by injection. This technique must be taught to the parents and child (if appropriate) so that the child can be given an emergency injection at home if needed. (More information is given in the Emergency Treatment Pack, Booklet No. 5.)

Surgical treatment

Surgical treatment of CAH is usually carried out when the child is about one year of age,

by which time she is big enough to be operated on safely, but not old enough to have become embarrassed about the appearance of her genitalia. The nature of surgery required depends on the degree of masculinisation. Usually the surgeon will need to reduce the size of the clitoris very carefully, preserving the delicate supply of nerves and blood vessels to the tip so that normal sexual relationships can be experienced in the future. Also, the surgeon may try to open the entrance to the vagina. The length of stay in hospital varies, but between 5 and 10 days is usual.

Sometimes it is not possible to open the vagina completely at the first operation and in some girls a future operation may be needed, optimally at the time of puberty and certainly well before they want to start a sexual relationship.

Once puberty has started, therefore, it is recommended that such girls are referred either back to the original surgeon, or to a gynaecologist, in order to reassess whether any further surgery is going to be needed. When surgery is needed in young adulthood, patient and parent support is advisable. This should be initiated long before surgery and should be through a psychologist with experience in this area.

IS STEROID TREATMENT HARMFUL?

Steroid treatment in CAH is ***VITAL*** if the child is to grow and develop normally. If the correct dose is used, there are no 'side-effects' whatsoever, as the treatment is merely replacing what the healthy adrenal cortex should normally produce. However, if too much hydrocortisone treatment is given, growth will be slowed down. Too little will result in excessive growth, and even signs of premature sexual development, due to increased male hormone production.

Too much fludrocortisone may cause high blood pressure but this is rarely a problem after the first few months of life. When it does occur, the high blood pressure is only temporary and corrects itself when the dose is reduced. Your specialist may therefore recommend regular blood pressure monitoring, especially over the first six months.

HOW MUCH TREATMENT IS NEEDED? WHAT TEST SHOULD BE DONE TO DECIDE THIS? HOW OFTEN SHOULD MY CHILD BE SEEN IN CLINIC?

There are no infallible rules for the correct dose of steroid treatment as this varies from patient to patient. In general, non-salt losers will require lower doses than saltlosers. As you would expect, each child will require a larger dose of hydrocortisone, or equivalent steroid, as he/she gets older and bigger, and the requirement may rise quite steeply at the time of puberty when growth is rapid.

Opinions vary concerning the place of blood and urine tests in CAH. Blood tests and blood pressure measurement can be used to estimate the dose of fludrocortisone required, while the growth rate and bone maturation, with or without blood/urine tests, can be used to estimate the hydrocortisone dosage. Practice varies between centres but, whatever the controversy, ***there is complete agreement that the most important test for determining the correct hydrocortisone dose in CAH is the growth rate.*** This can only be determined properly if the child is brought to the clinic regularly, three of four times a year, and this is of as much importance as the regular giving of the medicine.

A piece of information which is used in addition to the growth rate is the x-ray of the hand and wrist which tells us about bone development. This test, known as a 'bone age', is useful from roughly two years of age. If bone development is advancing too fast this usually means that a higher dose of hydrocortisone is required.

In summary, children with CAH should expect to attend the clinic three or four times a year to be measured, to have an increase in the dose of steroids roughly once a year, to have their blood pressure measured at each visit, and to have an x-ray of hand and wrist performed once (occasionally twice) a year. Additional tests of blood or urine are at the discretion of the centre concerned.

WILL MY CHILD GROW NORMALLY AND REACH A REASONABLE ADULT HEIGHT?

With careful treatment it should be possible for children with CAH to grow at a rate sufficient to achieve an adult height within the normal range. However, even with the very best treatment, it is not always possible to achieve perfect growth in CAH, and some individuals may end up shorter as adults than they would otherwise have been. Also, the dose of hydrocortisone required to control the CAH may make it difficult for some children to keep their weight down, and this can be a particular problem for girls at adolescence. It may be helpful to have the input of a dietician — you should discuss this with your specialist.

AT WHAT TIME OF DAY SHOULD TREATMENT BE GIVEN?

There is still some debate about the timing of treatment in CAH. Some centres recommend that the main dose of hydrocortisone be given at bedtime, while others believe that it should be given as early as possible in the morning. These differing views are probably related to the fact that every patient is different, and your specialist will discuss with you the exact type of treatment which best suits your child.

HOW DOES THE MANAGEMENT OF CAH CHANGE IN AN EMERGENCY?

As stated on page 5, hydrocortisone is a very important hormone to combat stress. Normally, individuals produce more cortisol than usual in response to illness, injury, surgery, etc., and it is very important that individuals with CAH, being unable to make this response, have their medicines increased at such times. Your specialist will advise you about what to do if your child is ill. In addition, the following guidelines for the management of illness in children with CAH may be useful.

Minor illnesses

If the child has a minor illness, such as a mild cold, but is otherwise completely well, then no increase in hydrocortisone is required.

Moderate illnesses

If the child is unwell, for example with a respiratory infection or tummy upset so that he/she is unable to continue normal activities (playing, going to school, etc.), it is advisable to **double** the usual daily dose of hydrocortisone, dividing this into two or three doses spread out over the day. Once the child has recovered, usually after two or three days, the usual dose of hydrocortisone can usually be restarted — contact your family doctor about this if in doubt. (Note: the extra hydrocortisone should be given **even** if the child is receiving treatment for the infection.)

Severe illnesses

In severe illness, especially when this is associated with diarrhoea or vomiting, the child needs hydrocortisone urgently and may be unable to absorb it when it is given by mouth. Under these circumstances it is essential to give an injection of hydrocortisone **without delay**, i.e. even before contacting your GP or the hospital. If the child seems very unwell, particularly if pale, clammy or not responding normally, the injection should be given at home by the parents. This should be done before contacting the GP or the hospital because delay may be very serious. Sometimes parents find it difficult to put their finger on why they think their child is ill and delay giving an injection because they don't want it to look as though they are over-reacting! If in doubt, it is always better to inject first and ask questions later, **remembering that no harm can come from giving an unnecessary hydrocortisone injection**. This is why you need to know how to give the injection. Speak to your specialist about this if you are unsure or have any questions. (Additional information is available in Booklet Series No. 5.)

To give the injection, the correct dose of hydrocortisone (25 mg for babies, 50 mg for children aged 1—5 years, 100mg for older children) should be drawn up into the syringe; the injection is given into the side of the thigh mid-way between the hip and the knee where there are no important structures that can be damaged. The injection is given

intramuscularly; the needle is put into its full length and the whole injection given. The child's condition usually improves very rapidly but a doctor should always be consulted if your child has been thought ill enough to require administration of a hydrocortisone injection. If symptoms persist, particularly vomiting, the child must be admitted to hospital for an intravenous drip to replace salt and water.

Surgery

Surgical procedures such as dental extraction, which need a general anaesthetic, should be carried out under hospital supervision and it will usually be necessary for your child to receive an injection of hydrocortisone before the anaesthetic. Extraction of teeth with local anaesthetic does not usually need any special precautions: however, you should tell your dentist of your child's condition.

Illness notification

To ensure good communications, especially in the rare instances where children and parents are unable to indicate that the child has CAH, it is important for children to carry notification of their condition, and the need for steroid treatment, on them — either a steroid card, MedicAlert necklace or bracelet (details of these can be supplied by your specialist or the Child Growth Foundation).

CAN MY CHILD RECEIVE LIVE VACCINATIONS DESPITE BEING ON STEROIDS?

Yes. The doses of steroids used in CAH are equivalent to the amount produced by the adrenal glands under normal conditions, and there is NO reason for your child to be managed differently where vaccinations are concerned. The situation is quite different for people who are receiving large doses of continuous steriods for different medical reasons, for example in the treatment of inflammatory conditions.

How CAN I GET ADVICE IN AN EMERGENCY?

As stated above, it is best to give extra hydrocortisone first and then ask for advice!

If, having given an emergency injection both you and your family doctor are uncertain what to do in a particular emergency, it is usually best to arrange for an ambulance to take the child to the **nearest** casualty department.

When you are on holiday and/or abroad make sure that you carry a letter from your specialist giving details of your child's regular treatment, as well as the treatment recommended for emergencies. Your consultant will advise you about what is needed for your child and what to take with you.

DOES CAH CAUSE PROBLEMS AND WHAT SUPPORT IS THERE FOR FAMILIES WITH CAH?

As stated at the beginning, the diagnosis of CAH often comes as a deep shock to parents and, since the condition affects the appearance of the genitals in girls, it is not surprising that families feel deeply distressed and confused, often finding it difficult to talk about the problem to their family and friends. It is also understandable that girls with the condition become upset by the need for genital examination and surgery, particularly if this has to carried out during adolescence. Much work is still needed to explore the feelings of both parents and patients with CAH so that proper counselling and support can be given. For the moment, we would simply reassure families that feelings of confusion and distress about CAH are both common and completely normal. Specialised psychological support will be available at your hospital and it is important that you discuss any anxieties, particularly for girls with CAH, with your specialist.

With respect to support groups for CAH, a national network is in operation so that there will be a co-ordinator within your area. The person who will put you in touch with your co-ordinator is Mrs Lesley Greene, Research Trust for Metabolic Disease in Children (RTMDC),Goldern Gates LodgeWeston Road, Crewe, Cheshire CWI IXN (telephone 01272 250221).

For some parents, it is particularly helpful to speak to or meet another family who have a similarly affected child with CAH. Ask your CAH support group co-ordinator, the CGF or your consultant about this.

Regarding financial support, some families have been successful in obtaining a Disability Living Allowance (previously Attendance Allowance) and this is well worth applying for. It is particularly likely to be granted for pre-school children who are more prone to acute illness, and thus require greater supervision.

CONCLUSION

CAH is a treatable condition where affected individuals lead normal lives and have a normal life expectancy. This is what one parent said, on reading this booklet:

"It might be worth ending on a encouraging note indicating that such children usually lead perfectly normal lives. We lived through the first years in the expectation we would have to care for a sickly child throughout most of his life. Nothing could be further from the truth. Our experience with Michael was that he became responsible enough to take his own cortisone at a very early age and we really don't have to worry about the day-to-day care."

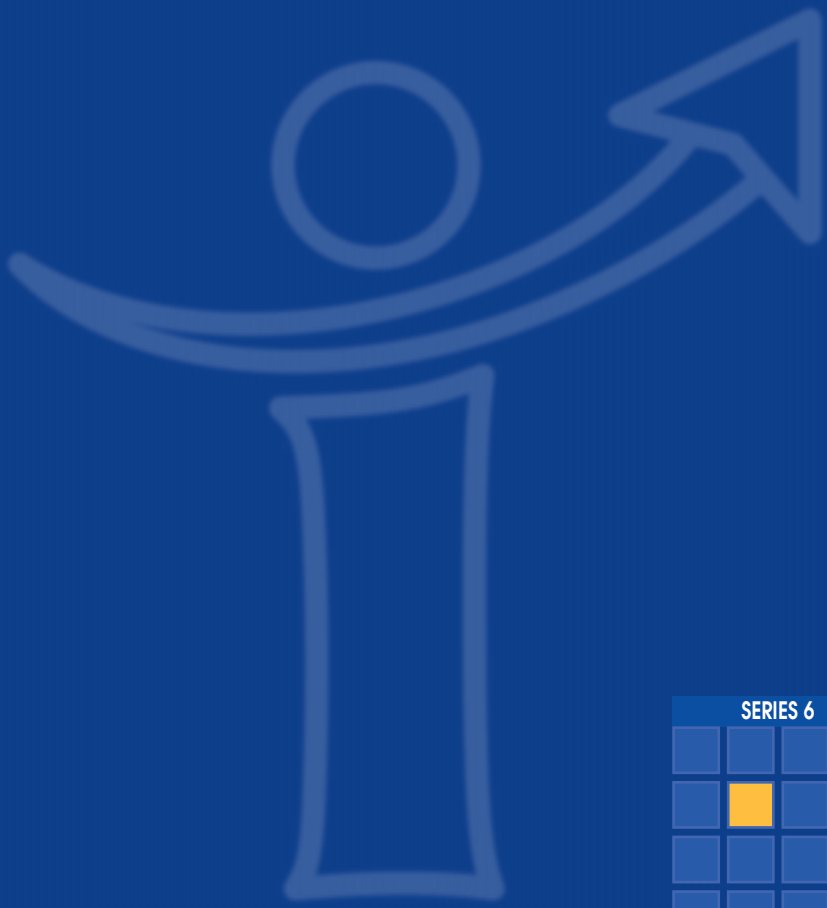
We thank Dr Peter Swift and Professor Ieuan Hughes, Lesley Green and Sue Elford from Research Trust for Metabolic Disease in Children, Irene McArthur and the parents from the CAH Support Group In Glasgow for their helpful comments and criticisms.

Further reading

Your Child with Congenital Adrenal Hyperplasia by G L Ware published by Educational Resources Centre, Royal Children's Hospital, Melbourne, Australia.

Emergency In formation Pack for Children with Cortisol and OH Deficiencies and those Experiencing Recurrent Hypoglycaemia (Booklet No. 5) in the series from the Child Growth Foundation/BSPED.

NOTES



SERIES 6

JULY 2004 **P060604**

serono