INFORMATION BOOKLET

FOR ADULTS

WITH

CONGENITAL

ADRENAL

HYPERPLASIA

Introduction.

This brief guide to the symptoms and treatment of the Congenital Adrenal Hyperplasia has been produced through discussion between support groups - the child growth foundation, the adrenal hyperplasia network and the congenital adrenal hyperplasia group and medical specialists - Dr Gerard Conway and Dr Richard Stanhope.

Although every effort is made to achieve accuracy, errors and the results of new research may invalidate some of the information.

Dr Gerard Conway
The Middlesex Hospital
Mortimer Street
London W1N 8AA

Support Groups:

Child Growth Foundation

2 Mayfield Avenue
Chiswick
London
W4 1PW

Tel: 0181 995 0257 / 994 7625
Fax: 0181 995 9075

E-mail: cgflondon@aol.com

Adrenal Hyperplasia Network

17 Newton Road
Lichfield
Staffordshire
WS13 7EF

Tel: 01543 252 961
Fax: 01543 411 761

E-mail: AHN@mlcull.demon.co.uk

Congenital Adrenal Hyperplasia Group

2 Windrush Close
Flitwick
Bedfordshire
MK45 1PX

Tel: 01525 717 536

E-mail: Sue@elford98.freeserve.co.uk

Congenital Adrenal Hyperplasia

Information for Adults

Growing up with a medical condition such as Congenital Adrenal Hyperplasia (CAH) can be very difficult and for some a lot of support is required. The problems of having CAH can be considerably reduced if the right type of information is made available and discussed sensitively. As young adults the issues of sexuality and fertility need early discussion so that adult life can be approached with confidence. These issues can be difficult for some parents, children and doctors.

As with any medical condition there are wide variations as to how each person is affected by CAH and not all that is in this leaflet will be relevant to everyone. For instance, CAH has different effects on women compared with men. This leaflet describes how some adults see the condition of CAH affecting their lives. Addressing these feelings enables them to find ways to deal with them. Further support may be found through the family, friends, a support group, a counsellor or a doctor. A specialist with a particular interest in the problems associated with CAH is needed at times.

What is Congenital Adrenal Hyperplasia?

The adrenal gland sits above each kidney – one on each side of the body. It is made up of a medulla (middle) which makes adrenaline – this part works perfectly normally in CAH. The outer part of the adrenal gland is the adrenal cortex which makes three main hormones called steroids. These steroids are secreted into the blood stream and are necessary for normal health. It is the adrenal cortex and its hormones which are involved in CAH.

The 3 main steroids involved in CAH are:

- ***Cortisol*** controls how the body copes with stress, emotional and physical, such as infection or injury. It also helps to control blood sugar levels, raising these levels if they become too low particularly in children.
- ***Aldosterone*** helps to regulate the salt levels in the body. Aldosterone causes the kidneys to conserve salt if there is too little salt in the diet, or if a lot of salt is lost due to excessive sweating. Conversely, if a lot of salt is eaten the adrenal cortex reduces the amount of aldosterone secreted, allowing the excess salt to be passed in the urine.
- ***Androgens*** are a group of male hormones, one of which is testosterone. For the rest of this booklet the term *testosterone* will be used to refer to the androgen group. Testosterone is produced by the adrenal cortex in both males and females and they control the formation of pubic hair at the onset of puberty. Testosterone is also produced by the testis and in small amounts by the ovary.

There are five main enzymes in the adrenal gland which convert cholesterol into the important steroid cortisol. If any of these enzymes are missing or defective then not enough cortisol is made for the needs of the body. The body, recognising the low levels of cortisol, will try to stimulate the adrenal cortex to make more by 'pushing' the gland harder by a stimulating hormone called ACTH made in the pituitary gland. The constant unsuccessful stimulation causes the cortex to increase in thickness and become 'hyperplastic'.

In the most common form of CAH there is a deficiency of an enzyme called 21 hydroxylase. In this form of CAH the production of cortisol and aldosterone is low while testosterone, the male hormone, is produced normally. As the body pushes the adrenal gland harder trying to correct the low cortisol level more and more testosterone is made. In boys, this excess of testosterone causes early sexual development. In females, the excess in testosterone can cause abnormal genital development before birth while in adults it causes irregular periods, unwanted hair growth and acne. Only by correcting the levels of cortisol with substitute therapy does the body recognise normal levels and stop producing excessive amounts of testosterone and the circulating levels become normal.

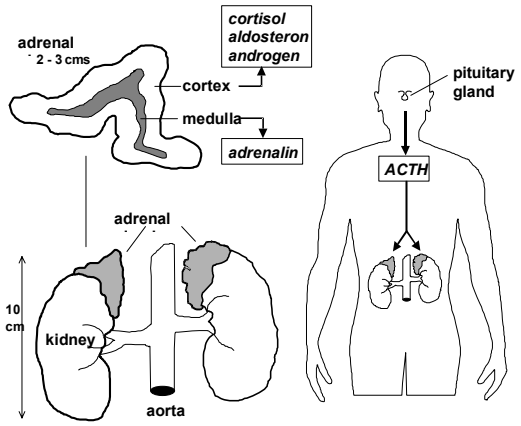
The hormone disturbances caused by CAH

- Lack of cortisol
- Lack of aldosterone
- Excessive androgens

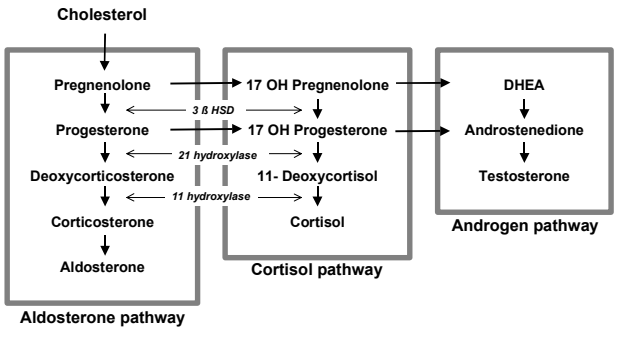
Different types of CAH

There are many different grades of severity of CAH depending on the degree of impairment of production of cortisol and aldosterone. In the most severe type of CAH aldosterone is completely lacking and loss of salt from the body is the most prominent problem – **salt losing CAH** accounts for 80% of children with CAH. The loss of salt in the urine is uncontrolled and can cause acute dehydration, very low blood pressure and vomiting. The levels of salt (sodium and chloride) and sugar (glucose) fall in the blood, and the potassium level rises. This is an 'Adrenal Crisis' needing very urgent treatment as a potentially life threatening condition.

In the 20% of children with less severe CAH, non salt-losing CAH, the salt balance is normal. In stressful situations, however, some people with non-salt losing CAH may become salt losers and need extra treatment. Girls born with **non salt-losing CAH** are usually healthy, but often born with an enlarged clitoris and the labia may be partially fused because of the excess of testosterone. In boys, **non salt-losing CAH** produces no detectable signs at birth and the diagnosis is made when the penis enlarges at a very early age along with early pubic hair and rapid growth in height the result of high levels of testosterone. These changes may not occur until 4



The pathways of steroid production in the adrenal gland. Cholesterol is taken up by the gland and three main groups of steroid are made - aldosterone, cortisol and androgens. Some of the enzymes which make the chemical changes are shown in italics - 21 hydroxylase is the most common enzyme to be affected in CAH. The steroid, 17 OH progesterone is just above this enzyme and therefore a useful marker of how the adrenal gland is working in CAH.



or 5 years old.

The mildest form of CAH – **late onset CAH** - affects women at any age. Symptoms of unwanted hair growth or irregular periods can start at any time after puberty. Often treatment with steroids is not necessary in women with **late onset CAH**. Instead, giving oestrogen as in the oral contraceptive can regulate testosterone from the ovary. In fact, the treatment of late onset CAH is usually the same as for the polycystic ovary syndrome because the two conditions are so similar. In men, late onset CAH usually goes unrecognised although it may cause the sperm count to be low.

All of the types of CAH above are deficiencies of the enzyme *21 hydroxylase* which account for over 90% of people with CAH. The next most common deficiency is of the enzyme *11-beta hydroxylase*. Treatment of this type of CAH is more complicated because high blood pressure can be severe if treatment inadequate. The balance of treatment in 11-beta hydroxylase deficiency is very difficult and an experienced specialist is essential. Deficiencies of other enzymes are exceedingly rare and beyond the scope of this pamphlet. A specialist will be able to offer more information on the rarer forms of CAH. *Adrenogenital syndrome* is an older name for CAH which is still used occasionally.

Treatment of CAH

People with CAH have a normal life expectancy and for most people there is very little interference in every day life if the condition is well managed. Adults with CAH require life long follow up in specialist hospital clinics in order to maintain the correct level of medical care and to provide early advice on fertility when needed.

It is advisable to have regular medical checks even if everything is well controlled. For women, checks should be made 2 - 3 times per year. For men, once per year is usually sufficient. The main aim of treatment is to maintain normal cortisol levels and control of salt loss, in those who are salt losers. In women, good management of CAH brings about control of testosterone levels, regular periods and improved fertility. In men, good management can reduce an aggressive behaviour and control excessive libido. Also, the sperm count can fall if CAH is not well treated and an increase in the dose of steroid treatment for some months can restore fertility in men.

Replacement of cortisol

The object of cortisol treatment in CAH is to replace inadequate production of cortisol by the adrenal glands allowing the body to recognise normal levels of cortisol in the body, which reduces the need for the adrenal gland to produce excessive amounts of testosterone.

How do doctors work out how much cortisol to take?

If the cortisol dosage is too low the adrenal gland will try and make more, but only succeed in making more testosterone. The effect of these excess androgens is growth of body hair in women and infertility in both sexes. **In severe forms of CAH, too little cortisol treatment may make the body unable to cope with stress and infection, so that major surgery or an illness like 'flu' can be life threatening.** If the dose of cortisol treatment is too low, a blood test will establish this by showing high levels of several adrenal hormones of which the most commonly measured are:

- 17 Hydroxyprogesterone
- Androstenedione
- Testosterone

Other side effects of steroid treatment are:
Headaches (sometimes associated with taking prednisolone and dexamethasone)
Feeling sick / nausea / tiredness
Finding it difficult to cope with stress
Water retention (sometimes associated with taking Hydrocortisone and Cortisone acetate)

There are four types of cortisol replacement treatment: Hydrocortisone, Cortisone acetete (now rarely available in the UK), Prednisolone and Dexamethasone. They vary in their dose and duration of action. Hydrocortisone is another name for cortisol. Prednisolone is 5 times more potent, and dexamethasone is 40 times more potent than cortisol. Both prednisolone and dexamethasone are comparatively long acting, where as cortisone acetate and hydrocortisone are shorter acting, and need to be taken 2 to 3 times a day. The dose in each dexamethasone tablet is not convenient for fine-tuning of treatment leading to a danger of taking too high a dose. Dexamethasone is established for use in pregnancy for women who are at risk of having a child with CAH. Hydrocortisone is also used as an injection at times of adrenal crisis or when vomiting prevents the tablets from being taken.

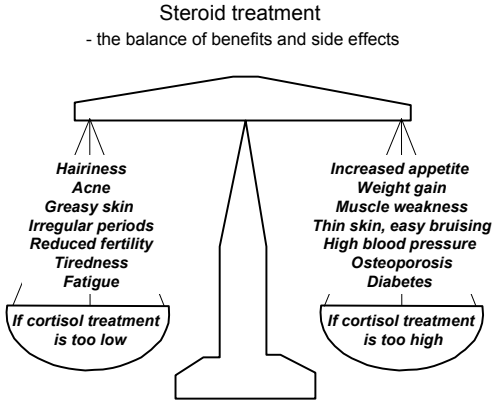
Types of steroid tablet and examples of daily doses

Type of steroid	Tablet sizes	Dose range for adults
Hydrocortisone	10 mg, 20 mg	10 – 40 mg
Prednisolone	No coating: 1mg, 5 mg Enteric coating: 2.5, 5 mg	2.5 – 10 mg
Dexamethasone	0.5 mg, 2 mg	0.1 – 0.75 mg

The exact dose needed each day for each individual will differ, due to the variation in body size, different rates of absorption in the bowel. It is important to assess the individual response, taking into account timing of the day that medication is taken. It is important to be clear how many tablets are missed so that the correct balance of dosage can be achieved. Many people find it difficult to take tablets regularly and this can lead to conflict with doctors.

At times of stress such as during illness or physical or emotional shock, the dose of cortisol replacement treatment should be increased in a controlled way. The exact regime should be agreed in prior discussion with you doctor. For example, a double or treble dose is usually advised at times of stress. The body increases cortisol in situations of high mental stress and it may therefore be necessary to give an increased dose even if only for one day for exams,a driving test, or a sudden bereavement for example. If an illness causes prolonged vomiting which prevents tablets from being taken, then an injection of hydrocortisone 100 mg should be given into the muscle and a doctor should be called. The technique of injection can be learnt by members of the family with instruction from the practice nurse.

Unless there has been substantial over dosing of fludrocortisone, blood pressure should not present a problem taking into account the wide variation in general blood pressure levels.



Replacement of aldosterone

All individuals with salt losing CAH need to replace the lack of aldosterone. Only one form of treatment is used here. Fludrocortisone is given in a dose of 50 – 300 micrograms. Too little fludrocortisone can cause low blood pressure and dizziness. Too much fludrocortisone can cause high blood pressure and headaches. The correct level of fludrocortisone is determined by measuring blood pressure, potassium and the salt sensitive hormone *renin* in the blood every few years.

Adrenal crisis

If there is a severe shortage of cortisol then an individual's life can be in danger with an adrenal crisis. This event is very rare is will never happen to most people with CAH who take treatment regularly, make appropriate increases in dose when ill and seek help early if unexpectedly unwell or vomiting. The symptoms are faintness, nausea, vomiting and abdominal pain. An adrenal crisis can be triggered by a stressful illness such as severe flu or gastro-enteritis. An injection of hydrocortisone should be given immediately. Admission to hospital may be needed so that fluids can be given into a vein – a '*saline drip'*. If in doubt – always give an injection of hydrocortisone and go to the nearest accident and emergency department.

Why not take the adrenal glands out?

There is an operation to remove the adrenal glands called *adrenalectomy*. People who have no adrenal glands, a condition called Addison's disease, require the same medication as those with CAH, so the question sometimes arises that if treatment is difficult for any reason, why not remove the adrenal gland? Adrenalectomy is not a minor operation and the long-term safety of this option has not been established. For some women in whom all fertility treatments have failed, then adrenalectomy might be the only answer.

Issues for adults with CAH – men and women

The problem of being over weight

Many people with CAH, women in particular, have a battle to avoid gaining weight. Mostly, weight gain in CAH is a side effect of treatment with hydrocortisone, prednisolone or dexamethasone and to a lesser extent, the oral contraceptive pill if used. Short stature and depression can also make weight gain more likely. Careful monitoring of weight is important as prevention of weight gain is better than a harsh weight reducing diet. Careful adjustment of treatment and attention to diet and exercise should enable most people with CAH to avoid obesity. A normal healthy diet which is low in sugar and fat and high in fibre is best. A sugar craving can be hormone driven although the effect of various treatments is unpredictable. Regular sustained exercise of an hour several times per week may be required to keep fitness levels up and weight down.

Will my baby get CAH?

It is very rare for a parent with CAH to pass the condition of to a child – less than 1 in 100 chance. CAH is autosomal recessive condition meaning that one copy of a defective gene must be inherited from each parent to make an individual who is *homozygous* for CAH. A parent with CAH has two defective genes but their partner is likely to have two normal copies that will make up for the defect. That is, children of a parent with CAH will have one defective gene and one normal gene. They will be *heterozygous* for CAH – a situation which has no effect on health. It is current practice to test the partners of people with CAH by measuring 17-hydroxyprogesterone in the blood rather than to have a genetic test. The chances of anyone with no symptoms or family history of CAH carrying a defective gene for 21-hydroxylase is between 1:50 and 1:100. The issue of genetic testing should be discussed with your specialist well before you are planning a pregnancy because some tests are useful if performed ahead of time. The advice on this topic is likely to change with time so specialist advice should be sought as part of pre-pregnancy counselling.

Who should have genetic testing for CAH?

A change in the genetic code of the gene that is responsible for making the enzyme 21-hydroxylase is the most common cause of CAH. There are only a few centres in the United Kingdom and elsewhere, where this genetic code can be 'deciphered' and the defect can be precisely defined. Knowledge of this genetic defect in CAH has practically no effect on treatment and so the tests are not performed routinely. The only time that a genetic test is useful is to diagnose CAH before birth so that the foetus can receive early treatment by giving dexamethasone to the mother. In this way, a masculinisation of the external genitalia of female foetus with CAH can hopefully be prevented. In practice, genetic testing and antenatal treatment is only useful for families with one child already affected by CAH when the chances of having another child with CAH is one in four.

Sports

There is no restriction in any type of exercise for individuals with CAH. Indeed exercise promotes good health and helps with both weight control and the prevention of osteoporosis. With strenuous exercise an extra dose of steroid treatment is advisable especially in the early stages of training.

For those who wish to enter competition sports, the International Olympic Committee recommends declaring CAH and that steroids are being taken for medical reasons. The IOC is unable to comment on whether hormone replacement steroids needed by those with CAH would affect the status as a competitor. They were not willing to be precise on drug testing procedures which would probably include testosterone and related compounds. They consider this an important issue, and with further investigations in the future, would try to establish the effect of medication necessary in CAH on the screening tests.

The Amateur Swimming Association can see no reason to exclude any one with CAH from competitive swimming, but recommended that the ASA should be informed of the competitor's condition prior to the event.

Travel

There should be no restriction for travel or recreation for individuals with CAH. If activity is likely to be stressful or if the destination does not have easy access to modern medical facilities, then is it wise to travel with hydrocortisone for injection. Vials of hydrocortisone for injection with syringes and needles should be taken and a dose of 100 mg given into the buttock in cases of trauma or severe vomiting. This action will make the situation safe until a doctor can be called. Supplies of hydrocortisone should be divided between hand luggage and other baggage to that in the case of one supply being lost or stolen there is always a back-up. A letter from a consultant can be useful so that contact can be made if illness occurs and to document the need for syringes and needles. Long-haul flights can be stressful and a double dose of steroids is advised.

Employment

In general there should be no restriction for employment for individuals with CAH. Commercial pilots, police, fire brigade and the armed forces are special cases where almost any condition requiring regular medical treatment can prevent employment.

Issues for women with CAH

For those women who find coping with the medical, surgical, psychological or sexual problems of CAH difficult, support and counselling should be made available. Sensitive discussion with specialists, general practitioners, fertility specialists, gynaecologists, counsellors, psychologists and CAH support groups can all be helpful. The difficulties experienced by some women with CAH arise because of decisions made many years ago when less was known about medical and surgical care. Young adults now should not have to face many of the problems addressed in this section.

Unwanted hair growth and acne

The high androgen levels that occur when cortisol treatment is too low, commonly cause problems for women with CAH. For many women acne and excess hair growth, 'hairiness' or *hirsutism* are the first features of high androgen levels. The aim of changing the dose of cortisol treatment is to reverse this process and it can be a very fine line between keeping free of unwanted hair and suffering the side effects of cortisol treatment - in particular weight gain. Because of these difficulties, many women with CAH require additional treatment for excess hair growth to oppose the action of androgens. The *'anti-androgen'* treatments available in the United Kingdom are cyproterone acetate, spironolactone, flutamide and finasteride. Cyproterone acetate is the only one of these to be in common usage and it works by competing with testosterone at the hair follicle to block the stimulation of hair growth. Anti-androgens are often given together with the combined oral contraceptive pill which reduces the amount of testosterone made by the ovary. Hirsutism takes several months to respond to treatment but cosmetic methods of treating excess hair such as waxing, shaving, creams or electrolysis are perfectly acceptable options which in no way cause hair to become thicker.

Contraception

There is no restriction for the use of different contraceptives for women with CAH. The choices of contraception include the sheath, the diaphragm, the coil and oral contraceptive pills. The combined oral contraceptive pill may have the added benefit of making irregular periods regular. Some women, however, find that the pill makes it more difficult to lose weight although a low dose pill might get around this problem. Dianette is commonly advised for women with CAH because it is particularly effective in suppressing unwanted hair growth. Progesterone only pills - 'mini-pills' can be used by women with CAH but they can cause difficulties with irregular periods.

Fertility

Most women with CAH have polycystic ovaries when pictured by an ultrasound machine. In fact, one in four of women without CAH also have polycystic ovaries, so this in itself is not a worrying feature though, of course, a concern to the individual. Polycystic ovaries are slightly larger than average and contain more small follicles, where the eggs develop, than average. Polycystic ovaries are associated with irregular periods and infertility which are both common in women with CAH. It may be that high levels of testosterone in childhood cause the development of polycystic ovaries in some women but this is far from certain. High levels of testosterone from the adrenal gland cause irregular periods and failure to produce an egg, *ovulation.* Irregular periods and failure to ovulate can improve with higher doses of cortisol treatment but the balance of treatment in this instance can be very difficult.

Irregular periods occur in about one third of women with non-salt losing CAH and one half of women with salt-losing CAH compared to one in ten of women without CAH. There is surprisingly little information about fertility in women with CAH and most of our knowledge on this subject comes from a time before modern fertility treatments. This 'historical' information says that two thirds of women with non-salt losing CAH are fertile without the need for treatment whereas only one in ten of women with salt-losing CAH are fertile. Most specialists feel that the prospects for fertility are better than these figures would indicate nowadays. If a woman with CAH has very irregular periods, then it is likely that specialist fertility treatment will be required. On the whole, fertility treatments will be the same as for women without CAH where a fertility tablet, clomiphene citrate, can be given. If tablet treatment is not successful then hormone injections (gonadotrophins, LH and FSH) can be used to bring about ovulation. If all of these treatments fail then *in vitro* fertilisation (IVF) might be needed but a specialist fertility clinic with experience in CAH should be consulted at this stage.

For women with late onset CAH fertility problems are usually less marked. In the mildest forms, steroid treatment might be necessary in order to conceive but it may be able to be stopped in pregnancy.

Pregnancy

Once pregnant, both the mother with CAH and her child should expect to be healthy in every way. There is discussion as to whether there should be a slight increase in dosage of cortisol replacement treatment in late pregnancy but high dose treatment should be avoided. The placenta protects the baby from any hormone imbalance in the mother and destroys any excess hydrocortisone in the circulation. In a recent review of 46 children born to mothers with CAH, all babies were healthy and normal. Two thirds of babies were delivered by caesarean section and only one third by vaginal delivery. There are many reasons why caesarean sections might be common in women with CAH but one worry is that earlier genital surgery might make normal labour difficult. Pain relief and the use of epidurals is the same in CAH as normal.

Gender Issues

Issues about gender often cause great concern but are not always addressed. It is important to understand that male and female gender identities are established before birth and not affected by hormone changes later in life.

Many women with CAH have successful heterosexual relationships, though for some finding a partner is difficult. There are some women with CAH who are more comfortable with bisexual and homosexual relationships but this may in some cases, have more to do with their concerns about the formation of their genitalia, with feelings of vulnerability and apprehension of a sexual relationship with men. The main point to remember is that if a woman has established heterosexual feelings, there is nothing about CAH that will change this, even if the testosterone levels rise from time to time.

In all women, testosterone levels can affect sex drive or *libido.* The effect of testosterone on libido however, is very unpredictable. Some women with CAH find that when testosterone levels are high, sex drive is increased. Reducing testosterone with higher dose of cortisol treatment can reduce excessive sex drive. A low libido is a more difficult problem as it is rarely improved by changing the dose of treatment – counselling might be an answer here.

The attitude of parents and doctors to gender and sex, and their own perceptions of this area, can influence how girls and women cope with CAH. Women appear to manage much better if, as children and young adults, their parents and doctors were able to discuss openly and sensitively, not just the medical side but also the emotional and sexual issues. This affects how women perceive their genitalia/bodies as acceptable to others.

Those who have been brought up with no information about the condition and are unable to talk to their parents or doctors, find it difficult to over come common barriers when problems are brushed aside and sexual matters are not considered as a matter for discussion. A young adult, who will probably have had vaginal surgery will find it hard to ask for advice. She may find it very difficult to be comfortable with her body, be anxious about the prospect of sexual activity and unable to see it as something pleasurable.

Genital surgery

All girls with CAH are born with a vagina, but sometimes it is difficult to establish the position or size as these can vary

considerably. The vagina may be covered with a small membrane and the clitoris is nearly always enlarged. Some girls with mild forms of CAH, do not need any surgery.

The timing of any surgery is dependent on the degree of virilisation and the feelings of the parents after discussion with their specialist as to the options available. In order to feel confident of decisions about surgery a second opinion may well be helpful. Surgery to reduce the size of the clitoris, a clitoroplasty, and to make a larger vagina, a vaginoplasty, can be done at any age and opinions vary as to the best timing.

Surgery on the clitoris is relatively straightforward. The aim of a clitoral reduction is to reduce the erectile tissue and preserve the nerves and blood vessels to the glans, the tip of the clitoris, so that it remains sensitive. Surgery required for the vagina and urethra in more complicated and varies considerably. If the vagina is positioned high near the bladder there can be the possibility of damage to the neck of the bladder, which controls the release of urine. Great care must be taken as any damage to the bladder could cause incontinence. Pelvic floor exercises can be of great help with any minor weakness with the bladder control, and will also help to strengthen the vaginal walls.

An examination just before the start of menstruation is often carried out to establish that the vagina is able to allow the use of tampons and also to make sure that the vagina will be big enough for comfortable sexual intercourse. Some specialists prefer to do this examination under a general anaesthetic, which though not strictly necessary, can make it less embarrassing for the young adult.

Some women may require surgery later in life for an enlarged clitoris or because of a small vaginal opening. It is rare for the clitoris to enlarge in adult life unless the control of CAH is very poor for a long period of time so that high testosterone levels cause the growth of the clitoris. Despite earlier surgery, the clitoris may remain relatively large in some women. In adulthood, vaginal surgery may be required to ensure comfortable sexual intercourse and to reinforce confidence in feeling feminine. Even if surgery on the vagina has taken place in childhood, it is not unusual for the opening of the vagina to be small. There are many surgical procedures using different methods to enlarge and line the vagina. 'McIndoe' techniques use skin grafts from the buttock of thigh to form a 'tube' like new vagina. Alternatively, a part of the colon can be transplanted to the vaginal space. Women needing vaginal enlargement should have information on the various techniques available and be allowed to discuss all the possibilities. For example, the two surgical techniques described above leave scars and increasingly experts feel that the less invasive use of dilators is best for women even for those who have a very short vagina. There are some surgeons who feel that vaginal surgery for an adult should only be considered if there is already an established relationship. The alternative view, often held by women themselves, is that early surgery is necessary for sufficient confidence and self esteem to establish a sexual relationship.

Surgery now has much improved over the past 30 years and the involvement of patients in decisions affecting their care has increased. Adults, who had surgery many years ago, may experience more difficulties from the effects of surgery, than those who have had more up to date operations. For instance, the practice of complete clitoral removal, clitorectomy, was once quite common. Alternatively, some erectile tissue of the clitoris might have been left secured under the skin in front of the vagina, this can be extremely painful during intercourse. Also, the glands that secrete the vaginal lubrication prior to and during intercourse might have been damaged making is necessary for a lubricant, such as KY jelly to be used for comfortable intercourse. Some women notice a build up of scar tissue after more than one operation on the vagina. This can cause lack of sensitivity within the vagina or pain during intercourse as the scar tissue is not pliable and does not stretch easily. Oestrogen cream applied to the vagina or the use of dilators can overcome some of these problems

In most women, a small vaginal opening or shortened vagina can be improved with dilators of various sizes that will allow successful sexual intercourse. By applying gentle but firm pressure to the vaginal opening by inserting a plastic dilator and, over a period of time, gradually introducing a larger size of the dilator, the size of the vagina will increase. Within months a significant increase in size can be achieved. For those women who are not sexually active, the vagina will tend to contract if dilators are not used regularly. It may be necessary to keep up a regular alternate day use of dilators for ten minutes or so. Some women feel less happy with the dilators and prefer to use their fingers. However, this method takes time, perseverance, motivation, and the ability to feel comfortable in touching the genital areas. Also, some women with CAH may have relatively small fingers; if there is a partner, their help may be of benefit until intercourse is comfortable. The use of a dildo may also be acceptable. Intercourse itself can gradually increase the size of the vagina but it takes an unusual degree of self-confidence to feel comfortable with this technique.

Some women find that everything to do with the mechanics of sex is very embarrassing. Teenagers, in particular, may find it difficult to find support and understanding if their parents find this uncomfortable territory. Extreme sensitivity is needed so that they can discuss how they can improve what they may see as an inadequacy of their body. An experienced sex therapist might be useful here. It must be remembered that women without CAH have genitalia in all shapes and sizes. The penis is a very adaptable organ which also varies greatly in size.

Orgasm

It may be that the surgical and psychological issues of sex make it difficult for women with CAH to achieve an orgasm. The complete removal of the clitoris, pain on intercourse, poor vaginal lubrication or lack of self-confidence may all contribute to a failure to achieve orgasm. A gynaecologist with specialist experience or sex therapist should be able to help overcome these problems.

Social concerns

As has already been said, those who have been able to have an open and informed relationship with their parents and doctors find the hurdles of CAH easier to overcome.

Adolescence is often a difficult time. If, added to this you have a condition which requires you to take regular medication, you may "different" and vulnerable making you unsure about relationships. Adolescent girls with CAH may need a lot of support. In all adults there are times when depression occasionally can lead to feelings of suicide. Some women with CAH feel that extreme mood swings are often caused by hormone imbalances and these concerns need careful, sensitive discussion with a specialist. For young adults and women, counselling can be of great help in establishing self-esteem and confidence and in developing strategies to cope with difficult situations that may be experienced with relationships. With a counsellor, it may be helpful to discuss and understand more about the condition. For many it will be the first time that they can talk openly about the issues of sexuality, how they perceive their bodies and female needs and how the individual sees the effect of CAH on their adult life

How and when to tell a partner about CAH can be a difficult problem. If you have confidence about your self it is much easier to put the condition into perspective for the person you are telling. Just because they now know you have CAH, this does not mean you have changed from the person they knew or loved before. This is not to be dismissive of the very real feelings a lot of women have about the fear of rejection from a potential partner. Feelings of being and looking different can put women off even trying to establish a relationship and may also cause overwhelming despair and depression. Some women find that talking to a counsellor who is not emotionally involved, can be of great help in generating confidence in herself.

The importance of adequate professional counselling has not really been addressed in many hospitals. Ideally it should be given by counsellors who understand CAH and the issues that arise from it. Always ask your doctor about counselling if you feel that it may be helpful. Some women find talking to others with CAH more helpful, as they feel they may well have experienced the same difficulties, but found ways to overcome them.

Issues for men with CAH

The problems of weight gain and the side effects of treatment can affect men as much as women. Short stature, which can result for difficulties in treatment of CAH in childhood, can be a particular psychological burden for men. High levels of testosterone in CAH can cause problems with aggression and excessive sex drive in men.

There are two important long term consequences of CAH which only affect men, both of which are the result of too little treatment with cortisol. These are infertility and testicular enlargement. If adrenal suppression is inadequate then the adrenal gland makes too much testosterone. High testosterone production from the adrenal gland can occur because the dose of cortisol is too low or because tablets have been missed. The testicles, which also make testosterone, then shut down as the testosterone levels are high. Unfortunately, 'shutting down' the testicles also means that sperm are not produced in normal quantities and this can cause infertility. Sperm production usually picks up if the adrenal cortisol treatment is increased allowing the testosterone production from the adrenal glands to fall, so the testicles can begin to work again. It can take several months the sperm count to rise after an increase in steroid treatment.

Testicular enlargement is a rare effect of under dosage of cortisol treatment and it takes many years of inadequate treatment for this to occur. Normal testicles contain a few adrenal cells that enlarge if the levels of cortisol in the body are low. This enlargement usually goes away if the dose of cortisol treatment is increased. Occasionally doctors advise a biopsy of the enlarged testicle to make sure that there is no hidden cancer. CAH does not cause cancer but it is reassuring to make this check.

Frequently asked questions about CAH

What should I do if I miss a steroid tablet?
If it is only a few hours since the dose was due, then take the treatment when you first think of it. If the next dose is nearly due then don't take a double dose - just leave the missing dose out.

What effect does drinking alcohol have in my treatment?
Moderate drinking of alcohol has no effect on CAH or its treatment. Large amounts of alcohol can affect how the liver breaks down steroids making treatment less effective. However, if you are drinking excessive amounts of alcohol then you have more to worry about than your CAH!

Can people with CAH be extra aggressive?
An excess of testosterone when CAH is under treated can lead to aggression. However, many things in life can contribute to aggressive behaviour including having a condition for which there is no cure and facing frustrations with relationships or with fertility problems. The advice of both a hormone specialist and a psychologist might be helpful here.

What are the long-term effects of steroid treatment?
In the modern era when the doses of steroids tend to be lower than in the past, no long term problems with steroid use are anticipated. If there have been times when high doses of steroids have been used then osteoporosis is about the only long term hidden problem. A measurement of bone mineral density can be made to test for this risk.

I seem to get a lot of infections - can this be part of CAH or its treatment?
Steroids - not CAH itself - can lower the effectiveness of the immune system. For instance, people who use high dose steroid treatment after an organ transplant are more prone to various infections. The standard doses of steroid treatment used in CAH should not have this effect but occasionally it seems that some people are particularly sensitive to one of the effects of steroids such as immunity. If frequent infections are a problem then specialist advice should be sought but doctors often have no easy answer to this problem.

I don't get on with my doctor, what should I do?
Your relationship with your doctor is personal thing - some work and some don't. It is worth while trying to find a doctor whom you find sensitive and understanding as you may need them for a long time. Try talking to friends or support group members about who they find helpful. Don't be afraid to try a few out or to get a second opinion. Also remember that there may be times when you need a technical expert who may not be the worlds best communicator.

Dr Gerard Conway
Department of Endocrinology
The Middlesex Hospital
Mortimer Street
London W1N 8AA

January 2000