BijnierNET

Information brochure for young people about

Congenital Adrenal Hyperplasia

Appendix to the Dutch 'Quality of Care Standard for Adrenal disorders'

Introduction

This information brochure is intended for children and young adults who have congenital adrenal hyperplasia (CAH) and also for their family and friends.

So when the authors of this brochure use the pronoun 'you', they are including those around you as well. Healthcare practitioners can also use this brochure as a source of useful information.

CAH is an hereditary condition; in the Netherlands it is called Adrenogenital Syndrome (or AGS). This brochure primarily uses the abbreviation CAH; however, the abbreviation AGS may be found when particular information is only available in Dutch.

You were probably diagnosed with CAH when you were still very young. This syndrome includes a group of congenital disorders of the adrenal glands. One of the enzymes that are necessary for the production of the stress hormone cortisol is missing from the adrenal gland. Your parents have always taken care of you. They took the decision about whether or not you will undergo an operation, whether you got extra hydrocortisone, whether family, friends and neighbours were told about your illness, and they will have spoken to your teacher at school.

Slowly but surely you are now taking more decisions yourself, and you are starting to stand on your own two feet. That's only natural, it's part of growing up. If CAH is treated effectively, you can lead a normal life.

The medical information in this brochure is based on the Dutch Quality of Care Standard for Adrenal Disorders and the 'Diagnosis and Treatment of Primary Adrenal Insufficiency: An Endocrine Society Clinical Practice Guideline' 2016 101 (2) 364 - 389. A Quality of Care standard describes what 'good care' means for people with a particular disorder or illness.

The intention of this brochure is to support you and your doctor/endocrinologist in that treatment. The brochure is not intended to replace the advice that your doctor gives you about treatment. If you have any questions, you should always discuss them with your own doctor.

If you are unable to understand any of the terms used in this brochure, you can download an explanatory list of terms from the alphabet on the AdrenalNET website ('The alphabet', only available in Dutch).

Say hello to ...
Karim aged 22

Karim has CAH. He is a student and is specializing in insurance law. He lives at home with two younger sisters and his mother is very worried about him. Alongside his study, Karim has always done odd jobs; he has been working as a taxi driver, doing evening and night shifts. At the moment he is working hard to complete his thesis and he has applied for a nice-sounding job. Karim wants to convince his parents that he can stand on his own two feet. His mother would prefer to keep him under her wing for a bit longer.

Transition

Transition means the transfer of a patient from the children's department (tailored to caring for you as a child) to the adult's department (tailored to caring for you as an adult). In order for this transition to be seamless, you need to start the process in good time. For example: by allowing the child to attend an appointment with the doctor by himself, or by allowing him to collect his own medication from the pharmacy.

Under Dutch law, the first change takes place when you reach the age of 16. As an adolescent, you then have the same rights and obligations as an adult. These are set out in the Dutch Medical Treatment Contract Act (Dutch only).

Information (Dutch only) about the rights and obligations of patients in the Netherlands.

In practical terms, this means that up to your 16th birthday, the paediatric endocrinologist will speak to you together with your parents, guardian or other competent adult. After your 16th birthday, the paediatric endocrinologist will speak to you alone. It will be up to you to decide whether you invite anyone else to be present, such as your parents or guardian. Further information about the Dutch situation can be found here (Dutch only).

The next change takes place when you become 18: treatment is then taken over by an endocrinologist in the adult's department of the hospital. You then effectively say goodbye to the familiar paediatric endocrinologist. In some cases, the paediatrician might feel that the transition should be postponed for a while. If so, he will discuss this with you.

In addition, you will be confronted with all sorts of other challenges in connection with school, your group of friends and the clubs where you play sport for example. From the age of 18 you will be eligible to vote, you might want to college or university in another city and you will be responsible for your own health and medical insurance. There are a lot of things for you to deal with, and it is important that you can go on doing the things that you want to do, without CAH putting any unnecessary obstacles in your way. And you can! You can do it. And - wherever possible - your healthcare providers will help you.

Getting back to CAH

CAH is a hereditary condition; in the Netherlands it is called Adrenogenital Syndrome (or AGS). It occurs in both boys and girls. There are many different forms of CAH, and the severity of the illness can differ from person to person. In many cases, CAH is detected when the heel prick is carried out soon after birth as part of the screening of newborn babies. If a baby has a more severe form of the disorder it will be difficult for nurses to determine the child's gender and this will immediately lead to further tests. But, in some cases, CAH is only detected at a later stage, for instance if a toddler is growing too quickly or there is excess hair growth.

CAH is hereditary

An error in the DNA, our genetic building blocks, means that too little of an enzyme (or none at all) is present in the adrenal glands. In 95% of cases, CAH is caused by a shortage of the enzyme known as 21-hydroxylase. The adrenal glands need this enzyme so that they can produce hormones.

The animated clip What is congenital adrenal hyperplasia (CAH)? explains what CAH is in simple terms.

Further information about the adrenal glands and the hormones that they produce can be found at adrenals.eu.

Or have a look at the theme page on CAH at het adrenogenitaal-syndroom.

Hormones send out signals

The shortage of the 21-hydroxylase enzyme results in a problem with the production of the hormones cortisol (stress hormone) and aldosterone (salt hormone) in the adrenal glands. Normally, the production of cortisol is precisely regulated. If the cortisol is too low, the pituitary gland (in the brain) produces ACTH, a stimulating hormone which encourages the adrenal glands to produce more cortisol. Once sufficient cortisol has been produced, a feedback signal is sent back to the pituitary gland and less ACTH is produced. If there is a shortage of the enzyme and too little cortisol is produced, the pituitary gland continues to produce too much ACTH.

The hormone ACTH goes on stimulating the adrenal glands because cortisol production hasn't increased. This results in the adrenal glands becoming enlarged (in medical terms, this is called hyperplasia) and they work even harder. But because of the enzyme problem they are only able to make one type of hormones, the male hormones: androgens. The end result is an excess of androgens. In addition, some of the ingredients for cortisol and aldosterone are piling up and these can have all sorts of effects on the body. The shortage of cortisol and aldosterone, and the excess of androgens, lead to a variety of symptoms. The severity of the disorder is closely linked to the enzyme deficiency.

CAH can give rise to:

- A shortage of cortisol (stress hormone);
- A shortage of aldosterone (salt-regulating hormone);
- An excess of androgens (male hormones).

Classic and non-classic CAH

CAH occurs in two forms: classic CAH whereby there is very little or no enzyme activity at all, and a milder form known as non-classic CAH whereby there is some but not enough enzyme activity. Two types of classic CAH are distinguished: with and without aldosterone deficiency. The type which has an aldosterone deficiency is also sometimes called 'salt-wasting' CAH.

Classic CAH (due to a 21 hydroxylase deficiency) occurs once in every 10 to 20 thousand births. Approximately 15 children are born with classic CAH in the Netherlands each year. Between 0.1 and 0.2% of the people in the Netherlands are affected by non-classic CAH.

With non-classic CAH, it is not usually a matter of a complete lack or a shortage of cortisol or aldosterone, but more often a matter of an increased production of male hormones. The excess of male hormones can cause problems, especially for girls.

The animated clip 'The HPA axis' on the Adrenals.eu website explains how our complex hormonal system works. CAH occurs in two forms. In the classic form there is no enzyme activity at all, or very little. In the non-classic form of CAH, there is still 30 to 50% of the normal enzyme activity. Two types of classic CAH are distinguished: with and without aldosterone deficiency.

How was your diagnosis made?

Most children with the classic form of CAH are diagnosed soon after birth. Boy babies are usually diagnosed when there is a positive result from the heel prick. Consequently, the disorder is detected early (usually one or two weeks after birth) and a life-threatening crisis - caused by the lack of cortisol and salt hormone - can be prevented. In the case of girls, CAH can be suspected directly after birth if their sex is not clearly obvious. If the CAH is less severe, the symptoms may not become evident until the child goes to primary school or even until puberty, and rapid growth or 'hairiness' raises doubts.

In all cases, doctors will measure the level of hormones in the blood, and also their various ingredients. Then a diagnosis can be made. CAH can be diagnosed with the aid of genetic testing. The results of the tests can also be important to other family members such as siblings.

How is CAH treated?

Although CAH cannot be cured, it is very treatable. The treatment depends mainly on the form of CAH that you have. The paragraphs below describe the treatment for both forms, as it will be provided after your 18th birthday.

Classic CAH

Treatment of classic CAH consists of a number of elements.
During a yearly or six-monthly check-up with the endocrinologist, he will do blood or saliva tests to make sure that the level of hormone substitution with hydrocortisone and/or fludrocortisone is sufficient. At the same time, he can check that the androgens are being suppressed enough. He will take your blood pressure and weigh you. Young and adult men will regularly undergo an ultrasound (echo) scan of the testes to check whether there are any small tumours in the testes.

Supplementing the shortage of cortisol
The shortage of cortisol is treated with medicines such as hydrocortisone. Hydrocortisone is often taken 2 or 3 times a day. It is important that you don't take too much (or too little) hydrocortisone, because both can be harmful to your body. Since healthy adrenal glands would normally produce extra cortisol in the event of illness, severe stress or an operation, people with this disorder need to increase the dosage of their medication in such circumstances to prevent a serious adrenal crisis. The endocrinologist will go through the stress instructions with you each time you visit the out-patient clinic. You can find them at website van BijnierNET.

Animated clip about the emergency injection.

Animated clip about an Addison crisis (also known as an adrenal crisis).

A new medicine has recently been introduced under the name Chronocort®. This medicine is slightly different from Acecort®. The tablets are taken twice a day: at 07.00 in the morning and at 23.00 in the evening. The hydrocortisone is released slowly into the small intestine. This has the advantage that the production of androgens is suppressed in the early hours (at around 04.00) and so it doesn't rise too high. This means that you will feel better and makes doctors think that, over the years, there will be less chance of developing associated illnesses.

Chronocort (trade name: Efmody) has been approved by the European Medicines Agency (EMA) since 27 May 2021, but the Dutch National Health Care Institute [Zorginstituut, ZIN] still has to approve its inclusion in the system which facilitates the refund of the cost of medicines; they still have to decide what proportion of the cost will be refundable and what cost - if any - patients will have to bear themselves.

At the time of publication (November 2021) it is still unclear when these medicines will be available in the Netherlands.

For the time being we are assuming that Efmody (this is the new name for Chronocort) will be available in the Netherlands from the second quarter of 2022 for AGS patients aged 12 and over. The product has been available in the UK and in Germany since September 2020.

Reducing the production of androgens

If you take hydrocortisone, this also reduces the production of androgens because the presence of hydrocortisone reduces the production of ACTH in the pituitary gland. ACTH levels in the blood are therefore lower, and the adrenal glands receive less stimulation to produce androgens. Hydrocortisone is often taken 2 or 3 times a day. Some patients also need to take the pill (an anti-androgen) as well, or need to have cosmetic treatment (e.g. shaving, waxing, laser treatment) to tackle the problems of excess hair growth.

Supplementing the shortage of aldosterone

The medicine Fludrace® (sometimes known as Florinef®) has the same effect as aldosterone in keeping the salt balance in our bodies at the right level. Over time, it can be necessary to adjust the dosage of fludrocortisone. This is normally done on the basis of blood pressure and blood tests.

Non-classic CAH

The treatment for patients with non-classic CAH is as follows:

- Children and young adults with the non-classic form of CAH can manifest early signs of puberty and have a higher bone age than their calendar age because of the increased male hormones. There is also a chance that they will ultimately grow to be less tall than parental heights would suggest. Treatment with hydrocortisone can then be considered in order to suppress the male hormones. However, the pros and cons of hydrocortisone treatment must be discussed fully with the parents and with you, even if you are still a child.
- On the whole, patients with non-classic CAH have sufficient cortisol and do not need hydrocortisone as maintenance therapy. Some patients, however, are unable to produce enough cortisol in stress situations. On the basis of a stimulation test (also known as a synachten test) the doctor will determine whether too little cortisol is being produced and will recommend that hydrocortisone is taken in the event of illness. The doctor will then discuss with you which stress schedule you should follow.
- Women who are found to be suffering from non-classic CAH (because they have excess hair, for example) do not usually need to be treated with hydrocortisone. But they may still experience symptoms because of the raised level of male hormones, such as skin acre, excess hair or menstrual and fertility problems. Besides cosmetic treatment for acne and excess hair, the treatment can consist of the pill (with or without anti-androgens), or - in more severe cases or in the event of fertility problems - hydrocortisone.

What else will the endocrinologist test during your visits to the out-patient clinic?

Unfortunately, CAH cannot be cured and that means that you will remain dependent on the medicines prescribed for you. Generally speaking, these medicines will help you well, but the use of medicines is always accompanied by a degree of risk. This applies to over-the-counter medicines and also to hydrocortisone and fludrocortisone. The endocrinologist will make a careful assessment of whether you are taking the right number of tablets and will go through the stress instructions with you. He will particularly check that you are not taking too much or too little medicine.
This is to prevent any long-term health problems. The dosage of medication is checked by means of blood and/or saliva tests. The risks of too much or too little medication are set out in the table below.

The endocrinologist and the nurse will also be on the lookout for psychological symptoms, because CAH might make you more likely to experience them. They will discuss their findings with you and see what form of care and support would suit you best.

Too much hydrocortisone

- Increase in weight
- High blood pressure
- High blood sugar levels, the start of diabetes
- Reduced bone density
- Increase abdominal fat
- Mood swings
- Severe tiredness

There can also be other symptoms and problems of too much cortisol, such as muscle weakness, bruising, changes in the body fat distribution (in the neck and around the waist), a full face, thin arms and legs, a weak and thin skin, and mood swings.

Too little hydrocortisone

- Adrenalcrisis
- Poor appetite
- Nausea
- Loss of weight
- Mood swings
- Acne, excess hair
- Menstrual problems
- Brown discoloration of the skin
- Enlargement of the adrenal gland (hyperplasia)
- Other adrenal tumours (giving rise to symptoms such as stomach ache or gastro-intestinal problems)
- Tumours of the testicles (TART)
- Severe tiredness

Too much fludrocortisone

- High blood pressure
- Fluid retention

Too little fludrocortisone

- Low blood pressure, dizziness when standing up
- Tiredness
- Salt craving

Which issues should you be particularly aware of?

At some time during your childhood it was discovered that you suffer from CAH. That is an important fact. Just like any other youngster, you probably want to lead the life of your own choice, and you certainly can. Nonetheless, we would like to take a moment to consider a few ' first times', whether you're 14 or 27 years old. There will be times in your life when the fact that you have CAH will have an impact on the decision you take. We know this from surveys of care providers, parents and young CAH patients themselves. They mention the following issues:

- Insight into CAH as an adult
- Leaving home to go to college or university
- Applying for a job and going to work
- Where you live, buying a house
- Leisure time, going out and holidays
- Relationships and sexuality
- Transport and travelling
- Sport and relaxation
- Finances and insurance
- Heredity, fertility and pregnancy

These points will be explained in the following paragraphs.

Insight into CAH as an adult

Sometime between your 16th and 25th birthday, you will be transferred from your familiar paediatrician to an endocrinologist in the adult out-patients department. This will coincide with a period of great change in your life; you might have just left home to go to college or just started working, you will be entering into new relationships and taking on more financial responsibility.

Points for attention:

- Encouraging independence and responsibility
  Parents or guardians usually take responsibility for the medical care of their children. As you get older, you will shoulder more and more of this responsibility yourself. This shift is just as important for parents and guardians as it is for you. How the changes take place differs from family to family and from person to person.

Because of that, you and the endocrinologist and/or the nurse specialist will assess: "What can you already do yourself and what needs a bit more time?"

- Increasing your insight into the disorder, the treatment and the consequences of CAH
  To make sure that you can take over responsibility for the care of your body, it is necessary that you know exactly what CAH is, why and how you are being treated, and what the possible consequences are. At this stage it is therefore important to continually refresh and update your knowledge.
  Read the brochure again from time to time and make a note of the questions you want to ask the doctor or nurse specialist.
- Preventing short and long-term complications (such as an adrenal crisis, risk of cardiovascular disorders, testicular tumours, reduced bone density)
  It is important that you follow the instructions for your medication closely. This includes possibly increasing the dosage of hydrocortisone if you are ill or are under severe stress for any reason. You need to be well aware of the stress instructions and know how to act if day-to-day life calls for it. Part of this is learning that you should never increase the stress medication too rapidly: too large a dose is not good for your overall health.
- Taking full part in the world around you so that the right steps can be taken to improve your situation if problems arise
  By 'the world around you' we mean school, your job, holidays, sport and so on. It is good to learn and discover, to get to know new people and to evolve as a person. Having CAH can sometimes get in the way, but your doctor or the nurse specialist can help you in this respect.
- The doctors and other professionals treating you will not only look at the medical side of your life, but also the social side. They do this so that they get a clear picture of your development and your hopes and wishes for the future (e.g. work, hobbies, sport).

Treatment team

Normally, the treatment of adults with CAH is in the hands of an endocrinologist, a nurse (or nurse specialist) and - if necessary - a gynaecologist, urologist or clinical geneticist.

These medical professionals will always try to approach you in a way that is appropriate to your age. What they advise will take your personal development, your personal circumstances and your own wishes into account. You can also introduce other issues you want to discuss with them.

If you notice that your appointments with the endocrinologist do not proceed as pleasantly as meetings with the paediatrician used too, then it is important that you make this known. If you feel unable to talk to the specialist himself about it, at least mention it to your parents. If you feel uncomfortable during your appointment, consider asking your parents to come with you next time, and then talk it out with the specialist. Perhaps you would be more comfortable with a different endocrinologist on the team. You have the right to choose your own doctor.

What can you count on?
The healthcare professionals will do their best to help you stand on your own two feet and to find answers to any questions you might have. In the background, your care is being transferred from the children's department to the adult out-patient clinic. But before that happens, you will be told the name of the usual person you will keep in touch with. Quite often, this will be a nurse or a nurse specialist. This person will explain what the procedure will be at the out-patient clinic. Your parents can also ask questions and get help and support if necessary.

More and more often, the initiative will be left to you. You will be helped by the fact that your knowledge about CAH is being regularly updated and extended. During the meetings with the doctor or nurse you can always discuss any matters that are important to you. You can decide whether or not to ask your parents to come to the appointment with you. Together with the nurse, you will set out a schedule, like a timeline, of what you want to have done or learned, and by when. In this way you will be standing up for yourself more and more and you will be better able to cope with the things that happen in everyday life. The better you can manage this, the better you have adjusted to the situation, the easier it will be to prevent health problems - both in the short term and the long term.

Leaving home to go to college or university

The switch from secondary school - which is usually quite close to the place where you live with your parents - to further education - perhaps in another town - can be a complicated process. There are so many things to think about, but that is part of the excitement.

Deciding that you want to live on campus or in lodgings and look after yourself is not quite as easy as it sounds. Up to this point, you parents have usually been able to make sure that you had sufficient cortisol in your blood. They have been able to recognize signs that suggest this might not be the case, and then they could talk to you about it. But that time will be over, and you will need to develop your own sixth sense in this respect.

Don't forget: the new course of study, a different study routine, the new people in your class and all the other new things in your life will all make demands on your energy. This is not to say that you will immediately need to adjust your dosage of hydrocortisone, but in the first few weeks of your new life it might be a good idea to keep a journal and make a note of how you are feeling from day to day. You can use the diary in the AdrenalAPP for this purpose. Discuss any queries or situations that arise with the nurse specialist. Perhaps some changes need to be made in the daily dose of hydrocortisone. You will be able to work out the best solution together.

Other than that, just carry on like any other new student. But you just need to pay a little more attention to how you are feeling.

Some students feel much more tired than usual, but help and support is available. Others might need a little more time to complete an exam paper.

The ECIO website provides useful information about further education for students with special needs. The informatie (in Dutch and English) relates to the situation in the Netherlands.

Applying for a job and going to work

Applying for a job

Although older employees more often suffer from chronic illness than younger employees, there is a steadily growing group of young working people with a chronic illness. According to the most recent estimates, just under half a million young adults (that's nearly 10 percent) suffer from a chronic illness during their youth. Thanks to good paediatric care, the vast majority (90 percent) of those youngsters grow up to become adults. About one quarter of young people under the age of 25 indicate that they have a chronic disorder, illness or handicap. Of those who have a job, around half say that this does not hinder them, the other half experiences problems - ranging from slight to severe - while doing their job.

Officially, when you apply for a job, you do not need to mention that you have a chronic illness and are dependent on medicines. And it is no longer necessary to have a physical examination when you apply for a new job.

In 2016, the Dutch Social Economic Council (SER) published a report under the title "Work: important for everyone", which contains many recommendations. These can be found under the following links:

SER's report and the Public Version of its recommendations

Talk to your endocrinologist about your chosen career whenever you feel ready to. CAH could play a role in your choice, although that isn't always the case. If you want to make sure, ask about it in good time. Even if you are not yet sure what you want to do. Discuss with your endocrinologist whether you should immediately mention that you have CAH, so that you can be prepared. Officially, employers are not allowed to ask about health matters, but in practice it does happen; there might be a gap in your resume (CV) for example.

Work

For most people, work is an important aspect of their lives. It gives you an identity. The first question you ask when you've just met someone is "Who are you?", closely followed by the second question "And what do you do?". What someone does, their job, helps you form an impression of that person. And exactly the same applies to someone with CAH.

As we said, CAH can be an issue with certain careers or jobs. If you want to become a pilot, for example, or join the army, there will be limits to what you can do. If the working conditions are extreme in any way, discuss with your endocrinologist whether you could reasonably pursue your chosen career. Sit down and talk about it, and get as much information as you can.

Shift work

Anyone who does shift work will know that you need time to adapt each time you change from one shift to another. That also applies if you have CAH. Your medication schedule will need to be adjusted as well. The nurse specialist can help you in this respect. Then it's just a question of trying it out and see how well you manage. It is best to find out whether you can cope during your probationary period in the first few weeks.

Where you live, buying a house

There are many issues to be dealt with if you want to buy a house, whether or not you have CAH.
In general, people who buy a house need to arrange a mortgage loan because they can't afford to pay for the house in one go. For a mortgage loan you need to go to a bank and quite often you are expected to arrange life insurance, either through the bank or an insurance company. This is necessary because if something was to happen to you, the loan would still have to be paid off by your family or partner.

Before life insurance can be arranged, you may have to have a medical examination. If the insurance company wants one, and you agree in writing, your

endocrinologist can provide you with a medical certificate. It is important to realize that the insurance company might impose extra conditions on the value of benefit for paying off the loan. This often leads to higher premiums or less favourable conditions.

Leisure time, going out and holidays

In principle, having CAH does not need to affect this side of life at all. Evenings out, student clubs or a pop festival: you can enjoy all these things. There is just one condition: make sure you take your medication in accordance with the prescribed schedule. If you don't and the carefully planned schedule starts to slide, your body will exact its own revenge!

Holidays outside the Netherlands also represent extra risk, and visits to countries with a greater chance of contracting 'traveller's diarrhoea' can be particularly risky. This is especially true if it is more difficult to obtain medical assistance because of distance or accessibility.

Travelling with adrenal insufficiency provides further details, but is only available in Dutch.

Going out

If you are planning a serious evening out, perhaps only getting home at 5 or 7 o'clock in the morning, don't forget to take your morning medication.
Set your alarm clock if you get home earlier! Your endocrinologist or the endocrinology nurse can give you further advice on this subject.

As one young lady put it: "The emergency kit is useful, but I can't tuck it into my bra"

Alcohol and drugs

These days, it seems to be common practice for everyone to drink alcohol and take drugs. These habits are not good for anyone's health, and certainly not for yours. Talk to your endocrinologist about what sensible steps you can take you to avoid problems. In any event, make sure that you carry visible evidence that you are dependent on hydrocortisone (such as an SOS card).

Buddy

it is advisable to make sure that you have a 'buddy', someone who knows that you have CAH and who knows what needs to be done if the cortisol level in your blood reduces and you therefore need to take hydrocortisone. People with CAH often say that when it gets to that point they are no longer able to assess

the situation for themselves. This is when your buddy needs to step in, when you react slowly or start acting in a strange way. It is therefore important that you explain the stress instructions to your buddy, so that he or she can help you avoid an adrenal crisis.

Relationships and sexuality

Some girls who have CAH will need to have an operation on their genitals. When girls reach the age of 12 they will have a final check-up from the paediatrician. Many young women indicate that - at the time - they found this a very intense time. That feeling is quite normal. It helps to know that the paediatrician understands that as well.

It is important that you have a good relationship with your paediatrician or endocrinologist, and that you are comfortable asking him or her questions about sex and intimacy. So don't avoid this awkward subject, OK? Even if you think you notice that the doctor is blushing ;-)

It is possible that the operation(s) will make stimulation or having sex difficult or even painful. Talk about this with your endocrinologist, who will then refer you to a gynaecologist.

In this situation, a boyfriend or regular partner can also develop a sixth sense and realize when you are suffering from a shortage of hydrocortisone. Or that you have taken too much. That's a good thing, too.

Transport and travelling

As a child you probably wore a SOS medallion, perhaps as a necklace or bracelet. Parents tell us that once children reach puberty they no longer want to wear the medallion because it makes it so obvious that they are dependent on hydrocortisone and they are therefore 'different'. That is very understandable. There's not much fun to it and you won't want to draw attention to your situation by wearing a medallion or having a SOS card on your seat belt strap. This is all very understandable.

Nonetheless, it becomes more important as you start travelling - more often and on your own - out of sight of the people who know and care about you. Neighbours and people at school will know about you. But if you are unexpectedly involved in an accident, for example, it may be complete strangers who need to act quickly. It is therefore sensible to carry an SOS card of some sort, but it's up to you.

Have a look at webshop of the Dutch Adrenal Society NVACP (in English) to see which alternative suits you best. For instance, there is a plastic SOS card you can carry in your purse or wallet. Another option is to have the AdrenalAPP on your mobile phone and put a sticker on the back of the phone.

If you are travelling, make sure that you yourself take measures to ensure that your dependency on medication is obvious in an emergency. Not only to yourself, but also to an ambulance crew, for example, who will only be able to treat you quickly and correctly if they have the necessary information. You are not helping them do their work properly if this information is not immediately visible. So just give it some thought and in this case make sure you are well prepared.

Sport and relaxation

People with CAH can usually play most forms of sport without problems. In the case of extreme physical exertion - running a marathon for example, or if there is a significant different change of altitude - you will need to take extra hydrocortisone. Talk to your endocrinologist or nurse specialist about how to adapt your medication schedule.

If you want to pursue sport at a highly competitive level, you will need a medical certificate from your endocrinologist in order to avoid problems in connection with drug testing. Take advice from the national association to which your club is affiliated, or from your national anti-doping authorities.

Finances and insurance

Standing on your own two feet also means that you have to balance your 'household budget'. From your 18th birthday, you will need health and medical insurance in your own name. You will have to pay monthly premiums for this insurance. In addition, like everyone else, you will have an 'own risk' (sometimes called 'excess') of EUR 385 a year. Quite a lot of money for someone who is still studying or has just started out on a career. Although you can easily switch to a different insurance provider each year, you will need a medical certificate if you want to take out supplementary health insurance. Health insurers are not obliged to accept requests for supplementary health insurance. This might mean that you are going to be faced with extra costs, so make sure you are fully informed.

Since 2020, as part of this 'own risk' system, patients in the Netherlands have had to pay part of the cost of 1 mg hydrocortisone and 62.5 µg fludrocortisone medication. These costs can amount to up to EUR 250 per year per insured person. Fortunately, many patients can claim a refund.

Until you are completely financially independent, your parents remain jointly responsible for your finances (up to 26 years of age if you are still in education or disabled, for example).

Own risk contributions and refunds
(Dutch only)

Heredity, fertility and pregnancy

If you have CAH and would like to have children, it is recommended that your partner is also tested to check for the presence of a mutation in the CAH gene. The chance that any partner is a carrier of the CAH mutation is roughly 1 in 50 (i.e. 2 percent). If you have CAH and your partner carries the CAH mutation, there is a much greater chance that any children will also have CAH. This chance is 50 percent. If your partner does not carry the CAH mutation, the chance that your children will have CAH is very small.

Both male and female patients who have classic CAH can experience fertility problems.
It is often found that women with CAH are also less fertile. This applies mainly to women with the classic form of salt-wasting CAH. If you don't take the medication according to the dosage schedule, this can have a negative effect on your fertility. In some cases it will be necessary to have treatment and counselling from a gynaecologist in order to become pregnant.

Men with CAH also have a chance of being less fertile. This can happen because your hormones are out of balance. Another reason can be that boys and men with CAH are rather prone to developing benign tumours in the testicles. These are known as TARTs: testicular adrenal rest tumours. They occur more often if your medication is not properly adjusted. Despite the fact that these tumours are benign, they can still block up the tubes that carry sperm or cause other damage. That is not something you want to happen.

If you have TART, it is advisable to have sperm tests carried out as a young adult (between the ages of 15 and 20, for example). Sperm can also be frozen to satisfy any future desire to have children, this is called cryopreservation. Unfortunately, there is no 100% guarantee that pregnancy will result from frozen sperm.

If you become pregnant, it is therefore important to get in touch with your endocrinologist as quickly as possible, and he will treat you together with a gynaecologist who has experience with CAH patients. The need for hydrocortisone can change during pregnancy, and the dosage will certainly need to be adjusted during the actual birth. In addition, your GP

will need to know what to do if an emergency arises during the pregnancy.

Dexamethasone during pregnancy

If a woman is pregnant with a child that might have CAH, prenatal treatment with dexamethasone is sometimes considered. The expectant mother will then take dexamethasone from the early months of pregnancy in order to treat the unborn baby. It is important that you are well informed about the pros and cons of this treatment, because there is still some uncertainty about the safety of this form of therapy. In the Netherlands, there are medical guidelines about the treatment. You should ask your endocrinologist for further information, or check out the BijnierNET website (Dutch only).

Extra preparations need to be made when you give birth as a CAH patient. The dosage of hydrocortisone needs to be increased in consultation with the endocrinologist. The Dutch Quality Of Care Standard For Adrenal Disorders includes a guideline for stress dosages around the time of birth. Together with the gynaecologist, you will draw up a plan for the birth. If you have had genital surgery in the past, you will also discuss whether a vaginal birth will be possible..

How is patient care organised?

The care provided to people with CAH is generally complex, because this is a rare disorder that calls for specific knowledge and expertise in terms of both research and treatment. The care plan involves a number of professionals, a multidisciplinary team, and this make careful organisation essential.

In the case of girls and women, attention is given to any problems which may ensue from surgical procedures carried out in the past. Cooperation with a gynaecologist, surgeon or urologist with specific CAH expertise is always worthwhile.

In the case of boys and men, attention is given to any tumours present in the testicles. The chance of such tumours developing increases if the CAH is not kept under control. Periodically, an echo (sonogram) will be made of the testes. For this purpose, the endocrinologist will refer you to the Radiology department.

Psychological support

At times, both men and women with CAH can feel sombre, depressed, angry, sad and anxious. These problems often arise because people find it difficult to come to terms with (internalize) their feelings and experience with the illness. Problems with your gender identity, sexuality and dissatisfaction with your appearance can also have an impact on your self-esteem. Psychological counselling would probably help, so don't hesitate to discuss such problems with your endocrinologist or your GP.

Dermatologist

Acne and excess hair can result from the over-production of androgens. Both can be treated by adjusting your medication, so that the body produces lower levels. Sometimes, however, this is not enough. Then cosmetic measures or other medicines will need to be taken, in consultation with the GP or the dermatologist. Cosmetic treatment options include hair removal by laser, epilation, creams or shaving.

Want to know more about CAH?

- www.bijniernet.nl
- www.adrenals.eu
- www.bijniervereniging-nvacp.nl

Membership of a patient association brings a lot of benefits, so membership is a serious option for you to consider. The patient organisations in the Netherlands also promote the interests of their members by entering into discussions about the care to be provided and the availability and supply of medicines. They also provide reliable information about the various clinical aspects of the disorders. In addition, they organise meetings so that patients can meet each other and/or learn about the latest developments. You also get a chance to meet other people of your own age who also have CAH.

Some healthcare insurers will pay part of the cost of membership of a patient organisation.

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The text of this brochure (Dutch and English versions alike) has been compiled with the greatest possible care. Despite this, they might contain errors. We request that you submit any additions or corrections to: info@bijniernet.nl

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