

# Congenital Adrenal Hyperplasia (CAH)



All rights reserved. No part of this book may be reproduced or transmitted in any form or by any means now known to be invented, electronic or mechanical, including photocopying, recording, or by any information storage or retrieval system without written permission from the authors or publisher, except for the brief inclusion of quotations in a review.



## For pricing and ordering information

C&W Bookstore  
Children's. & Women's Health Centre of BC  
Room K2-126, Ambulatory Care Building  
4480 Oak Street  
Vancouver, BC V6H 3V4  
tel: 604-875-2000 ext. 7644  
toll free: 1-800-311-1533 ext. 3  
fax: 604-875-3455  
email: [bookstore@cw.bc.ca](mailto:bookstore@cw.bc.ca)  
web: <http://bookstore.cw.b.ca>

## Acknowledgements

Developed by: Sheila Kelton, RN, BScN, Nurse Clinician

### Reviewers:

Dr. D. Metzger, Dr. J.P. Chanoine,  
Dr. L. Stewart, Dr. R. Rothstein,  
Sharon Connaughty, RN, MScN

Editor: Edna Durbach Ed.D, Dept. Patient/Family Education

Layout: Gail Soo Lum, Dept. of Patient/Family Education

Graphics: Piktografik



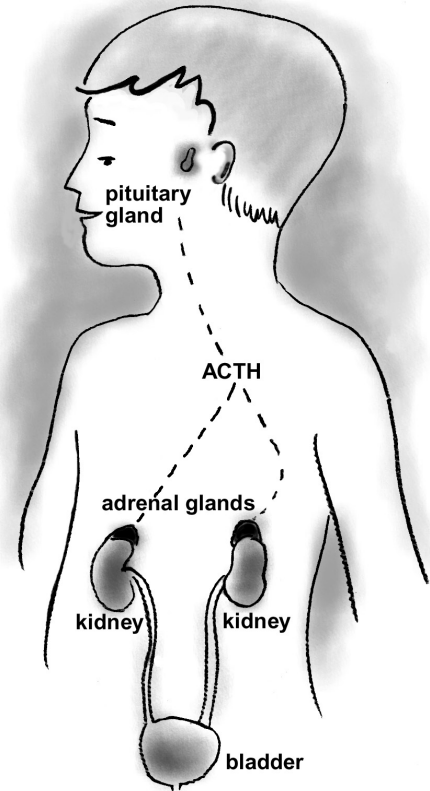
"When our son was an infant he was terribly sick. It was a scary time for us until the doctors realized he had CAH. They started him on his treatment and things soon looked up. He got better and so did we! A tiny pill three times a day is all it took."

Most parents are upset and worried when they first hear that their child has a health problem that will need daily medication for life. But you can take comfort in knowing that your child can develop and grow like any other healthy child. Congenital Adrenal Hyperplasia can be controlled. It takes only a daily replacement of adrenal hormones in the correct amounts and careful, ongoing, checkups. This book, and the health care team, will help you learn more about the condition.

## What is Congenital Adrenal Hyperplasia?

CAH is a life-long condition. The adrenal hormones are essential we cannot live without them.

About 1 in 15,000 babies is diagnosed with Congenital Adrenal Hyperplasia in infancy. Many more children have the milder form and are diagnosed later.



Congenital Adrenal Hyperplasia is an imbalance in the hormones produced by the adrenal glands. These glands lie on top of the kidneys. They play an important part in making several hormones. Children with CAH, have adrenal glands that are unable to make enough of the hormone cortisol. Sometimes, their adrenals are also not able to make enough aldosterone, for their needs.

## Resources for families affected by CAH

You can find these resources on the internet - note that website addresses may change. If you have trouble finding them, ask your health care professional or public librarian for help.

Giving an Intramuscular Injection:

<http://endodiab.bcchildrens.ca> and follow links to Endocrinology & Diabetes Unit For Families; Endocrine handouts.

Hydrocortisone replacement instructions

(English, Punjabi, Chinese and other languages):

<http://endodiab.bcchildrens.ca> and follow links to Endocrinology & Diabetes Unit For Families; Endocrine handouts.

Books or websites on CAH

"Congenital Adrenal Hyperplasia: A Parent's Guide"; C.Y. Hsu and Scott A. Rivkees, M.D.; AuthorHouse; 2006

[www.magicfoundation.org/www/docs/100/](http://www.magicfoundation.org/www/docs/100/)

[www.sickkids.ca/childphysiology/cpwp/Genital/GenitalIntro.htm](http://www.sickkids.ca/childphysiology/cpwp/Genital/GenitalIntro.htm)

[www.rch.org.au/cah\\_book/index.cfm?doc\\_id=1375](http://www.rch.org.au/cah_book/index.cfm?doc_id=1375)

[www.hopkinschildrens.org/cah](http://www.hopkinschildrens.org/cah)

[www.dshs.state.tx.us/newborn/hand\\_cah.shtm](http://www.dshs.state.tx.us/newborn/hand_cah.shtm)

Support Groups

Magic Foundation - [www.magicfoundation.org](http://www.magicfoundation.org)

CARES (Congenital Adrenal Hyperplasia Research, Education & Support) Foundation - [www.caresfoundation.org](http://www.caresfoundation.org)

Updated links are available from the Department of

Endocrinology at BC Children's Hospital

<http://endodiab.bcchildrens.ca>

\* website addresses may change

Most families feel themselves alone in the challenges they face when their baby is born with CAH. Some people feel sad or angry for what seems unfair. All these emotions are normal.

Most people say that after the first difficult months, they come to see that they are not alone. There are others out there who have been down the road before them. These families have helpful advice and support to offer. There are also a team of health professionals to respond to medical concerns. There is also the joy and excitement of watching your baby connect with you and the world around, to help you over the hard times.

We wish you well as you move forward in your life together.



**Cortisol** maintains the blood sugar in the normal range, keeps the blood pressure normal and assists the body to respond to any physical stress such as illness or injury.

**Aldosterone** prevents the body from losing salt. Without aldosterone, the kidneys remove too much salt and water and the baby becomes dehydrated.

Another aspect of the CAH condition relates to the hormones called **androgens**. They, too, are made by the adrenal glands. Androgens affect how the sexual organs develop before birth, and later, how the body matures at puberty. The adrenals of children with CAH produce too much androgen until the hormone balance is restored.

CAH ranges from mild to severe

- In its most severe form, the adrenals make almost no cortisol or aldosterone. This is called “*salt-wasting CAH*”.
- When the adrenals make enough aldosterone, but no cortisol, it is called “*simple-virilizing CAH*”.
- In the mildest form of CAH, the adrenals make enough cortisol, but only when they work “overtime”. In their effort to meet the body’s needs for cortisol they make too much androgen. The extra androgens trigger early puberty. It is often the signs of puberty, coming before it should, that calls attention to the problem. This form of CAH is called “*late-onset CAH*” because it shows itself only when the child is older.

You will understand the cause better if you think about it as happening in stages.

We need salt (sodium) to hold water in the body tissues.

Testosterone is a form of androgen.

## What is the cause of CAH?

Genes control the colour of hair, skin and eyes, the shape of the face and thousands of other functions of the body. The “gene blend” is often clear when you can see “father’s nose” and “mother’s eyes”.

- At its start, CAH is genetic disorder.

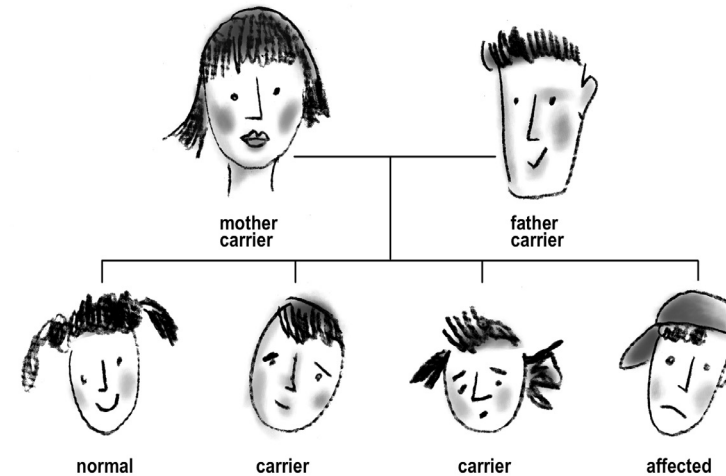
Genes contain the “pattern” for the person. From the moment when the sperm and egg unite, genes direct the baby’s development. Every human starts from two sets of genes – one carried by the sperm from the father and one held in the mother’s egg. The new human being is a “gene blend”. Some genes come from the father and some from the mother.

All of us carry some genes that are imperfect but they do not affect us. This is because, mostly, nature uses the normal gene from the pair at hand. The normal gene “dominates”.

CAH is an autosomal recessive gene disorder. It does not happen unless the baby inherits, from both parents, a gene deletion, that is, missing the same small part of the body’s pattern. The missing part is the instruction for making one of the chemicals in our body the adrenal enzyme 21-hydroxylase.

While both parents are carriers of this imperfect gene, neither parent has CAH or even knows s/he has an imperfect gene. This is because they both also have a normal dominant gene in their pair. The body takes direction from this normal gene.

This diagram shows the inheritance of CAH



### Q: Will my child with CAH have children with CAH?

A: The children of a child with CAH will all carry one CAH gene. But, the chance of their children having the condition is small. It will happen only if their partner also is carrying a CAH gene and the child then inherits the CAH gene from both parents.

It is a good idea to discuss your plans for future pregnancies with your endocrinologist or a geneticist (specialist in patterns of inheritance of genes). Research is testing medications that the mother can take, in the early stage of pregnancy, to decrease the “virilization” of affected female infants.

Each baby of parents who, like you, are carriers, has a 25% chance of having CAH; a 50% chance of being a carrier; and a 25% chance of carrying no CAH genes at all.

Our sexuality develops over many years. As parents, our job is to allow our children to find out about their sexual selves without shame. We must all try to “let them be” since nothing we do will change who they are. We can only help them learn self-respect. This is our challenge as parents of any child.

**Q: Will our other children have CAH?**

A: When both parents are carriers of a CAH gene deletion, there is a known chance of CAH for each child they have together. There is a greater chance, however, that other children will not have CAH. This is because each baby born inherits one gene from each parent so there are 3 possible gene blends:

- A baby could inherit the gene with the deletion from both parents – this baby will have CAH.
- A baby could inherit the complete healthy gene from both parents – this baby will carry only healthy genes.
- A baby could inherit one complete healthy gene and one gene with the deletion – this baby, like his parents, will not have CAH because the healthy gene will dominate. The child will, however, carry the CAH gene deletion.

- CAH is an enzyme deficiency

**The enzyme that is most often missing, or in short supply, is 21-hydroxylase. CAH is sometimes called enzyme 21-hydroxylase deficiency. Without 21-hydroxylase, the adrenals cannot make enough of some of the hormones they produce.** The lack of these hormones is the next stage in the cause of CAH.

The adrenals use cholesterol as their base material (main ingredient) for the hormones. By combining cholesterol with a series of different enzymes, the glands are able, step by step, to transform cholesterol into aldosterone, cortisol and androgen. By looking at the diagram on the next page, you can see the “production lines”. If the enzyme 21-hydroxylase is missing, or in short supply, the production of aldosterone and cortisol will be blocked.

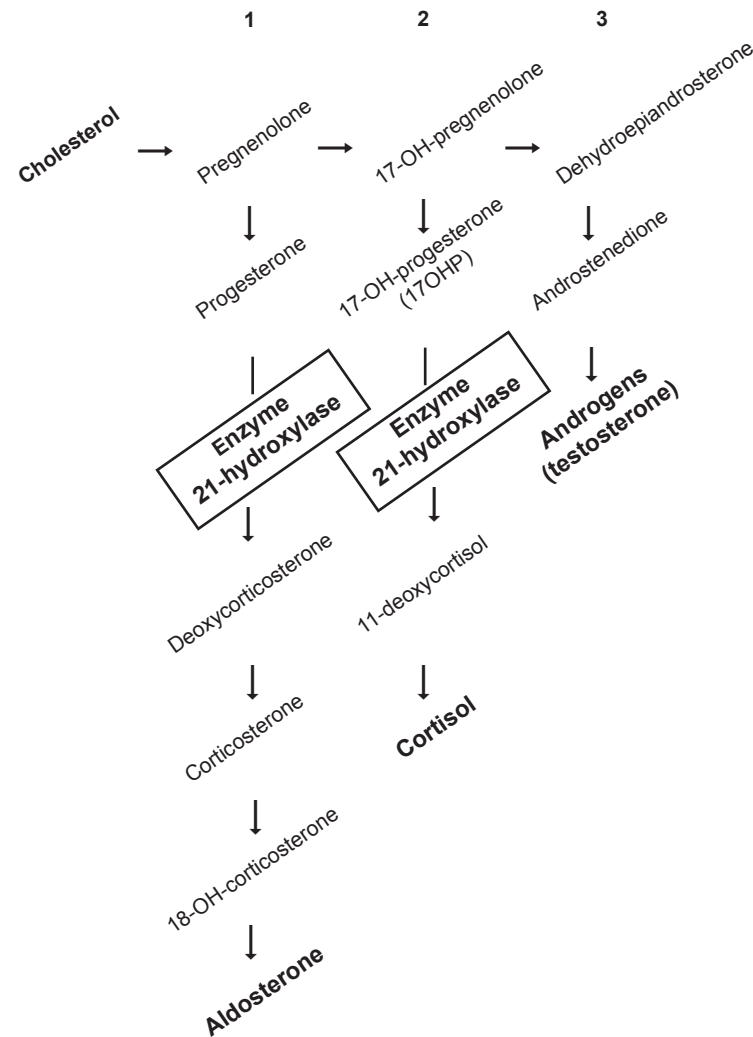
CAH is sometimes called 21-hydroxylase deficiency.

# The Making of Adrenal Hormones

Notice that:

- cholesterol has 3 separate conversion lines – one for each of the hormones it makes
- androgens, made in line 3, don't use 21-hydroxylase
- if cholesterol cannot be used in lines 1 or 2, it will all be used in line 3.

In a few children, it is not 21-hydroxylase deficiency enzyme that is missing but another enzyme. Your doctor will help you understand the specific nature of your child's condition.



## Q: Is there a cure for CAH?

A: At this time there is no cure. No one has yet discovered a way to make the body produce its own balanced supply of cortisol. But, the missing hormones can be replaced as described in this booklet.

## Q: Will my child develop normally?

A: Yes. Your child will grow and develop like any other healthy child if the missing hormone(s) are replaced in the right dose. His lifespan will also be normal.

## Q: My daughter prefers “boy toys and sports” to those most girls choose. Does that mean she thinks and feels like a boy?

A: In the past, we expected all girls to be interested only in what our culture decided were “feminine” activities. We expected boys to like only those toys and games society labelled “male”. We have come to realize that this tight box, into which we have put men and women, does not fit the great variety of personalities. We now understand that we can feel comfortable in our male body yet want to dance rather than play ice hockey. We can feel comfortable as a woman yet enjoy the thrill of soccer. Studies show that many girls with CAH do prefer sports and toys that we think about as “boys’ toys”. But, this would only be cause for worry if these girls felt like boys or were unhappy with being a girl. These studies do not suggest that these girls feel more like boys.



*A baby can go in to an adrenal crisis in an hour or two, while a school age child can be sick for many hours before reaching a crisis.*

**Q: What should we do if we miss a dose of glucocorticoid?**

A: Give the dose as soon as you remember. If you do not remember until it is time for the next pill, give a double dose. If your child is missing doses often, discuss this with your health care professional. As the child gets older, he may be able take a stronger type of glucocorticoid less often.

**Q: How quickly can my child go into an adrenal crisis?**

A: There is no standard answer to this. How quickly it happens depends on:

- . the age of the child (babies move into crisis much faster than older children);
- . the severity of the condition (children with salt-wasting CAH will go into crisis faster);
- . the amount of stress to the body (a sudden high fever is a greater stress than a mild cold).

**Q: Does my child need more hydrocortisone for emotional stress like exams?**

A: Most doctors will say no. Feelings do not challenge the body in the same way as physical illness or injury. Talk to your health care professional if you are concerned about stress in your child's life.

■ *CAH is a hormone imbalance*

The final stage of the cause of CAH is **the imbalance in the levels of adrenal hormones that comes about as the glands try to supply the body's needs.**

The health of the body depends on many things including a fine balance of the levels (amounts) of various hormones. The control centre for maintaining the balance is a small gland in the centre of the brain called the pituitary gland. This gland acts rather like a thermostat that turns the heating furnace (adrenal glands) on and off to control the heat (hormone supply) in the house (body). The pituitary gland turns the supply on by releasing the hormone ACTH (**AdrenoCorticoTropic Hormone**), into the blood.

Imagine that you have a house with 3 rooms (let's call them cortisol, aldosterone and androgen), with heat supplied by the same furnace (adrenal glands). The furnace is working fine but a series of levers (enzymes) must open the duct between the furnace and the rooms. One of these levers (enzyme) is broken. The duct is closed shut or can open only a little. Rooms cortisol and aldosterone need this duct open in order to get their supply of heat. These rooms have little or no heat coming in so the temperature falls. The thermostat (pituitary gland) registers this (releases ACTH) and the furnace (adrenals) starts working to change gas (cholesterol) into heat (hormones). The furnace starts cranking out lots of heat. Even so, little to no heat gets through to where it is needed.

*When there is not enough heat the thermostat turns the furnace on so more heat is made. When there is not enough cortisol or aldosterone, the pituitary gland releases ACTH. This starts the adrenals working to transform cholesterol into more of the adrenal hormones.*

The cholesterol that cannot convert to cortisol and aldosterone is shunted into the making of androgens. In a female infant the excess androgens push the genital development toward a male form (virilization). If the imbalance is not corrected it triggers early puberty.

But, the duct that leads from the furnace, to room androgen, has no block. It is wide open. The result is that this room is flooded with heat. It is getting not just its own supply but heat intended for the other two rooms as well. It gets far too hot in here. There is now an unhealthy imbalance of temperatures in this house (body). It is far too cold in some rooms (not enough of hormones cortisol and aldosterone) and far too hot in the other (far too much of the other hormone - androgen).



Dissolve the measured amount in 1 or 2 teaspoons of sterile water, formula or breast milk. Divide the daily salt dose into 3 – 4 parts. Give a dose before feeds using an oral syringe. The salt dose does not have to be measured perfectly. Make sure baby has enough salt but if he has a little too much it will not do harm.

**Q: Am I giving my child a harmful steroid?**

A: Glucocorticoids are steroids but the amount your child is taking will replace what the body normally makes for itself. They will help rather than harm. There are seldom side effects. Some people take very large doses of steroids to control conditions like asthma. In large doses, steroids have some unwanted effects.

**Q: Can my child have a sleepover or go to camp?**

A: We encourage you to treat this child as any other. Just make sure that the responsible adults know about the medicines, and how to increase the dose of hydrocortisone if your child is ill or hurt. If the camp is far away from health care, an adult needs to know how and when to give injectable hydrocortisone in an emergency.

**Q: Should my child receive the regular immunizations?**

A: Yes. Even though your child is no more likely to get infections than other children, managing illness is always a challenge. So, many doctors suggest that children with CAH also be immunized against flu, chicken pox, pneumococcus, and meningitis. If the immunization causes a fever, give the "illness dose" of hydrocortisone.

*It is a good idea for your child to have a flu shot each year.*

## Questions from parents

Do not put the dose of medicine into the bottle. If baby will not take the whole bottle she will miss part of the dose.

### **Q: How do I give a pill to a baby?**

A: Crush the correct dose with a pill crusher. Dissolve it in 1/2 teaspoon of breast milk or formula. Draw the mixture up in an oral syringe (a special syringe with a soft tip and no needle. Put the syringe into the front of baby's mouth. As baby begins to suck, push the plunger.

One form of glucocorticoid (Pediapred®) comes in a liquid form. It is measured and given using an oral syringe. It is not a good idea to have your pharmacy make the pills into a liquid form for you. It may not come in the right strength or hold its strength.

### **Q: How do I give salt to a breast-fed newborn?**

A: Discuss how much salt your baby needs with your doctor. Most doctors suggest 1-2 gram(s) per day. Here are 3 examples of how to measure and give salt:

- 1 gm = 6 ml of a 14.6% saline solution (ready prepared salt in sterile water available over the counter at the pharmacy).
- 1 gm = slightly more than a packet of salt (the kind on the table of fast food restaurants). Before you use salt packets for the first time, ask your pharmacist to weigh a few for you. Not all companies use 1gm salt packets. Dissolve 1 ¼packets in 1 or 2 teaspoons of sterile water, formula or breastmilk.
- 1 gm = slightly more than 1/8th teaspoon table salt.

This imbalance of hormones is the last stage in the cause of Congenital Adrenal Hyperplasia. In a female infant, the excess androgens will cause her genitals to “virilize” during fetal development. Her genitals will then look different from other baby girls.

## How is CAH diagnosed?

- All Canadian provinces and all the states of the USA take small samples of blood from all newborns to screen for certain conditions. Some include CAH as part of the screening.
- A newborn may seem well but within days start to feed poorly, vomit and **become dehydrated and very sick**. When there is no obvious reason testing is begun for CAH.
- Newborn girls are often diagnosed before they become sick because they are born with what is called “**ambiguous genitalia**”. This is a medical term which means the external genitals (sex organs) look different. When further checks are done, we find that their internal organs - upper vagina, uterus (womb) and ovaries are normal.

These tests of the blood may show very high levels of 17-OHP (see diagram on page 11). The tests suggest CAH although they will not pick up the milder forms.

The lack of cortisol and aldosterone means that the body cannot keep its salts and minerals at a healthy level and that the kidneys remove too much water from the body.

"The hardest thing for us to deal with in the early months was the way her genitals looked. It was really hard in the first day or two until we knew the sex for sure and could answer the question everyone asked: 'girl or boy?' It was really hard to stop our own questions about the future from crowding our heads in the next weeks. Now I wish I had just been able to set aside the fear and relax into the love I felt." Look in the section called "Special issues for girls" for some more information.

Boy and girl babies' genitals look the same in the first 2 months of development in the uterus. They have a small bud that will change into a clitoris or penis. They have folds that will separate into labia or fuse to form a scrotum. The line down the middle of the scrotum marks the fusion.

The genitals start to change when the boy's testes begin to make androgen (testosterone) around the 2<sup>nd</sup> - 3<sup>rd</sup> month. Large amounts of androgens start the growth of male genitals. High levels of androgens are being made by the adrenals of both boys and girls with CAH even before birth. This causes the clitoris to enlarge more than is usual and the labia to fuse some, or all, of the way, even in girls.

- In the mildest forms of CAH, the first signs come only in later childhood when **puberty starts too early**.

## What are the special issues for boys with CAH?

When they reach their teens, boys with CAH are likely to stop growing sooner than others. Their final height is slightly less than the other boys in their family. Some young men with CAH find their shorter height difficult. It affects how they feel about themselves. This is because our society values "tall" as something that makes men desirable. Parents who are prepared, can try to persuade their son to value other qualities.

Here is a time to be a sensitive parent! Ask your daughter whether or not she wants you with her when she goes to the gynecologist. This may be the time when she wants to be on her own. Some youth also want to talk about their sexuality without their mother.

### Adolescence:

During these years your daughter will come to understand her own body more fully. The changes that happen with puberty are another chance for you to further your child's understanding of CAH. This is an excellent time to go over, in some detail, what the hormones do in the body and how the medication replaces what her body cannot make for itself. As soon as your child shows signs of being able to take on self-care, start to give it over to her.

One important physical change is the start of menstruation. The vagina needs to be large enough so menstrual blood can flow out. Also, your daughter may want to use a tampon. Later she will want to know that sexual intercourse will not cause discomfort. If vaginal development is not complete, she will need to see a gynecologist. Most young girls are very nervous about this visit. You can ask the gynecologist for a "talking visit" first, with no examination. This gives your daughter a chance to feel comfortable with the new doctor. Discuss the timing of this visit with your daughter and your health care team.

Sexuality is on the minds of all adolescents. It will raise new issues for your daughter. Many parents find it hard to be open and relaxed for talks about sex with their youth. If you find you are too uneasy, do ask the health care team for some help. Your daughter needs to know there are adults she can go to if she wants a full and honest discussion about her sexual self and congenital adrenal hyperplasia.

## Testing for CAH

### ▪ Blood Tests

Blood tests measure the hormones of the adrenal pathway. The hormones that are made before the "enzyme block" will be in the blood at levels higher than normal. (Remember the furnace is working overtime.) The hormones after the "block" will be at low levels.

Blood tests will also measure the sodium level to show if there is the right amount of the salt-retaining hormone, aldosterone.

### Normal Blood Test Values\*

	SI Units (Canada, Europe)	Conventional Units (USA)
Cortisol (morning measurement)	275-690 nmol/L	10-25 mcg/dl
17-OHP (newborn)	<150 nmol/L	<5000 ng/dl
17-OHP (1mo – puberty)	<3 nmol/L	<100 ng/dl
Androstenedione (pre-puberty)	<1.8 nmol/L	<0.5 ng/ml
Testosterone (1mo – puberty)	<0.6 nmol/L	<17 ng/dl
Sodium	136-145 mmol/L	136-145 mEq/L

< means "less than" so, the normal 17 OHP level of a newborn is less than 150 nmol/L

\*varies depending on method, age of child, and time of day.

Your endocrinologist (a doctor who specializes in hormonal conditions) will help you understand the test results.

- An **ultrasound** of the adrenals and the pelvic area. This may show the *hyperplasia* – enlarged adrenal glands. They enlarge when they work overtime to make cortisol. The ultrasound can also show the uterus in a baby girl.
- **Chromosome check**  
Chromosomes hold the genes. Chromosomes may be checked if the infant has genitals that are different. This is because some conditions that result in different genitals may result from disorders in the chromosomes. The doctor may want to rule these out as part of the diagnosis for CAH.

## How is CAH treated?

The aim of the treatment is to match as closely as possible the hormone levels that normal adrenal glands would make. To achieve this, the dose will change as the baby gets bigger.

These life-saving glucocorticoids cost less than \$20.00 Canadian per month.

CAH is treated by replacing the hormone(s) that the body is not able to make for itself. Cortisol is replaced by one of a family of medications called glucocorticoids. The generic names of the glucocorticoids are: Hydrocortisone, Injectable Hydrocortisone, Prednisolone, Prednisone, Methylprednisolone, Dexamethasone. The trade names vary. *Hydrocortisone* is the *glucocorticoid* doctors most often suggest for children. So, we will use the word hydrocortisone to refer to the medication, although your endocrinologist may have suggested a different glucocorticoid for your child.

### Childhood:

During the school years, you can expect to have a healthy daughter. Keep in mind that her sense of herself as a sexual person starts to form in these years. Your response to her body is one of the key ways she learns about it. How you talk about the differences in her body will influence how she regards herself. For example, when she or her sister start to notice the difference in their genitals you can say something like:

*“Yes, these parts are not exactly the same – do we have any parts that are exactly the same? Let’s all look at our belly buttons. Are they the same? No, they are all different although they are all still belly buttons. Same and different isn’t like better or worse. We can feel good about how we are different and we can feel good about how we are the same.”*

We suggest that you use your child’s comments about how her body looks to start explaining her condition to her. You will need to go over this many times, each time adding a little more information. Reassure her that she is going to grow into a woman that can do, and be, whatever she would like. Tell her that at some time when she is older, she will see a special woman’s doctor (gynecologist) to make sure that her vagina is a good size.

Children with CAH almost always realize that their difference is not a casual one. If you brush aside their questions or comments, you take away a real chance to help them understand and accept their own body. If you never mention the difference in her body, your child may come to sense that the subject is a taboo. She will not feel she can come to you with her fears or concerns.

It is often hard for us, and young girls are no different, to have genital exams. Many parents ask the doctor to limit genital exams to only what must be done. You can also ask that the same doctor do the exam each time it must be done. In this way you protect your child from unnecessary focus on her genitals.

The genital exam is just a quick look at the external genitalia. It is not an “internal” exam.

There is some support, from medical studies, for the view that vaginal surgery, done in infancy, must be repeated when the child is older. Health professionals no longer feel there is clear evidence to say with certainty when is the best time for the surgery. This means that parents have a much bigger responsibility when making the decisions around surgery. They need to explore the topic thoroughly - ask your doctor for the most recent studies. There is plenty time to weigh the benefits of the earlier and the later surgery, or no surgery at all.



*"My Mom puts EMLA® cream on my arm an hour before the blood test. It numbs a little patch on my arm so the poke doesn't hurt."*

The second hormone, aldosterone, is replaced with fludrocortisone, Florinef®. As well, infants with salt-wasting CAH need to have a salt solution until they can salt their food to taste.

For the first 6 months the doctor will need to see the results of blood tests, done monthly, to find the correct dose. After that, the tests are done every 3-6 months. These tests can be done at a laboratory close to your home and then sent to the endocrinologist.

## Managing CAH when your child is ill or injured

All children get sick from time to time. Children with CAH are not sick any more than others. But, when children with CAH are sick, they need special care. Make an illness care plan with your health care professional.

When we are sick or injured, our adrenal glands make larger amounts of cortisol. The extra cortisol helps the body cope with the physical stress. But, since the adrenals of the child with CAH cannot respond to increase the cortisol level, he must take a larger dose of hydrocortisone until the body recovers. Deciding on the best hydrocortisone dose becomes a challenge when the child is sick or injured.

The general “rule of thumb” is:

- double the usual dosage for mild illness such as a slight fever or cold or an injury that requires stitches;
- triple the usual dosage for illness with a fever over 38.5°C or 101°F or if the child is in very bad pain for more than a few minutes. A major injury may need to be treated as a medical emergency with injected hydrocortisone.

Examples:

Nicholas is 14 months old. At noon he refuses any food or drink and becomes cranky. His mother notices his red cheeks. She checks his temperature and it is 102°F (39°C). She gives him a triple dose of hydrocortisone right away even though his next dose is due at 3.00 p.m. Her next step is to take Nicholas to the doctor who diagnoses an ear infection and starts him on a course of antibiotics.

virilization). The labia are joined so they look more like a scrotum, and the clitoris is enlarged, looking more like a penis. Girls with CAH have healthy ovaries, and a uterus, but sometimes the lower part of the vagina is not fully formed.

This is a very hard time for most parents. They are overcome with mixed feelings when they expected to feel only joy. Keep reminding yourselves that this condition can and will be treated. Keep your attention on cuddling, feeding and caring for your baby so that the bonds between you grow strong.

Infancy:

The main concern of many parents during the first year of life is “diaper distress”. Every time the diaper is changed the parent feels anxious about the genitals. In the past, the surgery to reduce the size of the clitoris and open the fused labia was done in the first year of life. This is still sometimes the case. However, unless the urine is not draining out easily, there is no hurry to do this surgery.

Until very recently health professionals believed that surgery made the genitals “normal”. Now, some women with CAH question whether doing the surgery in infancy was the right choice. They say that, as a result of the surgery, they have lost genital sensation and are not able to feel sexual pleasure. They feel that their genitals still do not look normal.

Surgery to reduce the size of the clitoris is called clitoral reduction. Surgery to open fused labia is called vulvoplasty. Surgery to create a larger vagina is called vaginoplasty. These surgeries do not have to be done at the same time.



Children with CAH should wear a Medic Alert bracelet. In an emergency this gives healthcare professionals the information they need about the child.

Older children can also go into an adrenal crisis but it does not happen as often. Older children are better able to let you know that they are feeling sick so you can give the extra doses of hydrocortisone. But, older children may have injuries like a bad fall that results in a broken bone or bad bleed. Over a period of time this can lead to an adrenal crisis. Inject hydrocortisone after a bad injury, call for emergency help, and be sure to tell the health care professionals about the condition and what you have done.

There is another situation in which a child with CAH may need hydrocortisone by injection - before a general anesthetic. Make sure the anesthetist knows about your child's condition.

## What are the special issues for girls with CAH?

### *Newborn:*

The early genital development of girls with CAH is disturbed because of the oversupply of androgens that their adrenals make. The adrenals start making androgens long before birth. These hormones force the genitals toward the male pattern. As a result they look different from what is expected at birth. Some look a little different - a clitoris larger than is usual and some joining of the labia ("lips" around the vagina). In severe CAH they look quite masculine (called

The doctor suggests she continues to give a triple dose of hydrocortisone every 8 hours. She follows the doctor's advice on ways to bring the temperature down. She makes sure Nicholas takes enough fluid to prevent dehydration. This means giving him small drinks every 20 minutes or so because he won't take a full bottle. Within two days Nicholas is getting better. Mother reduces the hydrocortisone dose to a double dose every 8 hours. When he is well again she returns to the normal hydrocortisone dose but continues the antibiotics until he has the full course.

Jennifer is 4 years old. She goes to a daycare where the staff knows about her CAH. They are aware that she needs special attention when sick or injured. She wakes one morning with a runny nose but seems otherwise well. There is no sign of fever. Dad decides to take her to daycare but asks the staff to watch her and call if she seems sick. When Jennifer complains later that she has a sore throat, the staff call Dad. He comes to take her home within the hour. He takes her temperature and finds it above normal. Jennifer lies down on the couch - unusual for this active little girl. Dad gives her a triple dose of hydrocortisone right away. He decides not to call the doctor because coughs and colds are common in the daycare. He continues the triple dosing every 8 hours and keeps her home the next day. The following day she is feeling better so he gives a double dose every 8 hours. On the third day she is quite well except for a little runny nose. Dad returns to normal dosing of hydrocortisone and Jennifer is eager to return to daycare.

There are no side effects for a short-term increase of hydrocortisone. If the child takes extra doses for longer than one week, decrease the dose slowly over 3 or 4 days.

## Managing CAH when your child is ill or injured

When the body needs a large amount of cortisol to deal with a major physical stress and none is available, the condition is called an adrenal crisis. The blood sugar and the blood pressure will start to drop. The body can go into a state of shock. Signs of this are weakness and vomiting, drowsiness that becomes loss of consciousness.

An adrenal crisis is a medical emergency. It is life threatening.

Keep injection supplies with you at home and when travelling. Know how to give the hydrocortisone injection yourself in an emergency.

Vomiting or diarrhea is a special challenge for children with CAH. They need extra cortisol at these times but they cannot hold the hydrocortisone in the digestive tract long enough for it to be absorbed into the body's cells. The situation can become a life-threatening *adrenal crisis*.

**The way to prevent the crisis is to inject (rather than swallow) hydrocortisone.** Most parents learn how to give these injections. Your health care professional can teach you. There are also instructions on a website listed at the end of this book. Some parents prefer to take their child to the hospital for the injection, but you should always be prepared to give the injection yourself in an emergency. You may not always be able to reach a doctor or nurse quickly enough, for example, during a long flight, when camping, or during snowstorms.

Infants can become very sick, very quickly. At the first signs of sickness watch your baby very closely. Set your alarm clock so you can check during the night as well. Give extra fluids, check temperature, give the extra doses of hydrocortisone if needed. **If your baby starts vomiting ("burping up" or "spitting up" is not vomiting) inject the hydrocortisone then contact your doctor or take your child to an emergency department.**



It is sometimes difficult to decide whether the baby needs an increased dosage. Call your health care professional to discuss the situation, but if you are unable to contact anyone, give the illness dosage just-in-case.

The dose of injectable hydrocortisone is large but does not cause any side effects. The body uses it up in 6-8 hours. After that you must start giving the pills again. But, if the child can't hold pills down, give another injection and go to an emergency department for further assessment. Keep in mind that the injection prevents the adrenal crisis but does nothing to treat the illness itself. Your child may need treatment for that as well.