The Leo Fung Center for CAH and Disorders of Sex Development

Congenital Adrenal Hyperplasia in the Newborn

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Introduction

This handbook will provide you and your family information about **congenital adrenal hyperplasia (CAH)**. While this guide will not answer all of your questions, it will provide basic medical facts that will help you to talk to your doctors.

It is important to know that CAH cannot be cured but it can be treated. Your child will need to take medicine for the rest of his or her life. If your child takes this medicine, he or she should have a completely normal life in every way.

Successful treatment requires teamwork between you and your doctor. The doctor will monitor your child in order to know what dose of medicine is needed. We ask that you give your baby the medication on the schedule recommended by your doctor.

Your family is not alone. The Leo Fung Center for CAH and Disorders of Sex Development (DSD) at University of Minnesota Amplatz Children's Hospital, provides a large network of support, including medical specialists, therapists and counselors who all have expertise in caring for patients with CAH.

What is congenital adrenal hyperplasia?

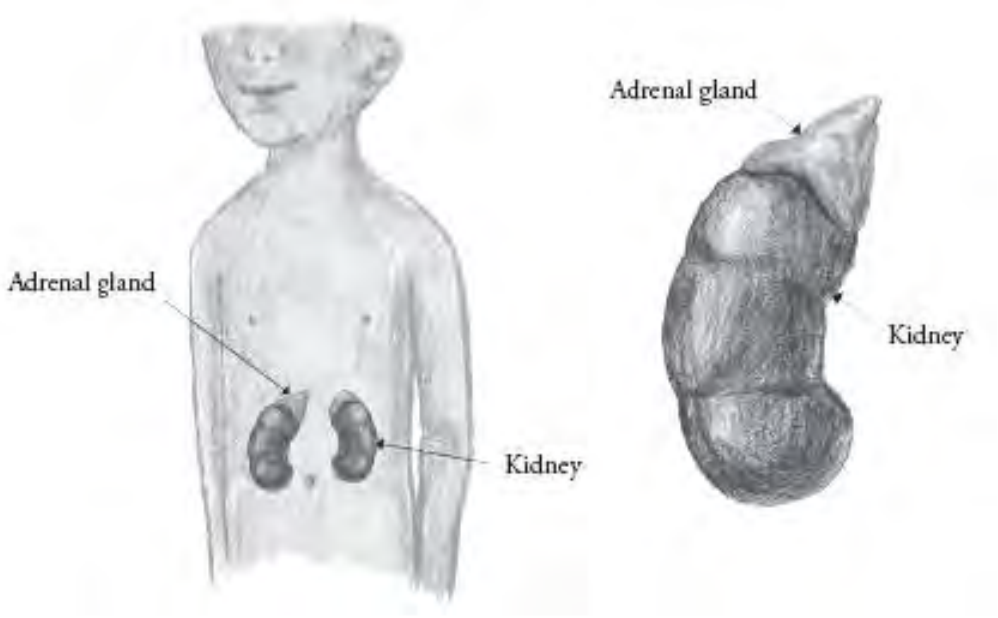
Let's begin by examining each word.

- **Congenital** means existing at birth (inherited).

- **Adrenal** means that the adrenal glands are involved. These glands are located above the kidneys. The adrenal glands produce three important hormones - cortisol, aldosterone and androgen. Each of these hormones has an important job in the body.

- **Hyperplasia** means an abnormal increase in the number of cells that make up an organ or tissue. This causes the organ or tissue to enlarge.

Congenital adrenal hyperplasia, then, is an inherited disorder that affects the production of certain hormones and causes the adrenal glands to become too big (hyperplastic).



Hormones involved in CAH

Cortisol (stress hormone): Helps control blood pressure, blood sugar and heart function. The body uses more cortisol during times of stress, injury and infection. Not having enough cortisol can be life threatening because it can lead to shock (dangerously low blood pressure), which is also known as an "adrenal crisis."

Aldosterone (salt-saving hormone): Helps balance water, sodium and potassium in the body. Without enough aldosterone, the body can't hold on to sodium and water.

Androgens (male hormones, such as testosterone): Both males and females have androgens. A male fetus needs androgens for normal genital development.
However, a female fetus should not have androgens or her genitals may not form normally (they may become more male in appearance).

Types of CAH

CAH may be “classical” or “non-classical.” Your doctor will tell you which form of CAH your child has.

Classical CAH

In classical CAH, the body produces more androgens (male hormones) than it needs. At the same time, there is too little cortisol (stress hormone) and sometimes too little aldosterone (salt saving hormone). This type of CAH occurs in about 1 out of every 15,000 births.

There are two forms of classical CAH: salt-wasting and simple virilizing.

- **Salt-wasting CAH** is the more common—and severe—form. With salt-wasting CAH, too much sodium and water are lost through urine, and the amount of potassium in the body increases, causing dehydration (loss of fluids) and very low blood pressure.
- **Simple-virilizing CAH** does not cause the body to lose sodium and water. Therefore, it is less severe than salt-wasting CAH. Like salt wasters, simple virilizers produce too many androgens.

Both salt-wasting and simple-virilizing CAH patients may develop an “adrenal crisis” during periods of physical stress (illness, surgery or trauma). This is a life-threatening situation and urgent medical care is needed.

Diagnosis in girls:

In girls, both kinds of classical CAH tend to be detected at birth because the genitals may not look normal. (Often they look more like boys’ genitals.) This is because their adrenal glands produced too many androgens (male hormones) in the womb. A typical female fetus does not produce this many androgens.

Diagnosis in boys:

In the male fetus, the testes already produce androgens. So if a few more androgens come from the adrenal glands, the genitals may look only slightly different at birth. (The scrotum may be more brownish in color, and the penis may be a little larger). Newborn screening is of particular value in boys, because they have no outward signs of the disease, and yet are at risk of "adrenal crisis" which can be prevented by early diagnosis and medical treatment.

Non-classical CAH

Non-classical CAH (NC-CAH) is milder than classical CAH. It is often referred to as "late-onset" CAH, because symptoms do not appear until later in life. Currently, this type of CAH is not detected through newborn screening of infants.

Newborns with NC-CAH do not have genital changes. Instead, the disease is diagnosed when the effects of excess androgen appear in childhood (rapid growth, early puberty) or during the teenage or adult years (too much face and body hair, severe acne, irregular periods).

Diagnosing CAH in newborns

Most babies with classical CAH will be identified through Newborn Screening. All states, including Minnesota, perform Newborn Screening for CAH. This simple blood test is done when the baby is 24 to 48 hours old.

Your doctor may recommend other tests (hormones, electrolytes) to confirm the Newborn Screening result of CAH. Later, your doctor may recommend genetic (DNA) tests. This requires another blood test. Family members may have this test as well. In fact, it is important for both male and female family members to be tested if they are thinking about having a baby. The genetic test for CAH may provide information that is useful for the doctor in caring for the baby's health.

Treating CAH

Treatment involves replacing hormones that the body cannot produce itself and keeping the body from making too much of other hormones. This means your child will need to take medicine regularly for the rest of his or her life.

While this appears simple, long-term success requires teamwork between your family and your doctors. You will need to:

- Make sure that your child takes the medicine faithfully.
- Keep all appointments with your child's doctors.
- See a pediatric endocrinologist (a children's doctor who specializes in hormones) to make sure the medicine is working.

Because each person is different, treatment is tailored to each patient. Your child will need to take medicine two to three times a day. This will ensure that your child maintains normal energy levels, the right balance of sodium and water, and normal growth and development.

Medicines

There are two kinds of medicines used to treat CAH. These medicines are steroids. They replace the hormones that are not produced by the adrenal glands. Your doctor will tell you which medicines are right for your child. They will need to be taken every day.

Children who have classical CAH need extra steroids during periods of increased physical stress. (Emotional stress does not require extra medicine.) The extra dose of steroids is called a stress dose. It can range from two to three times the normal daily dose depending on the severity of the stress to the body.

Ask your doctor how much medicine to give in a stress dose. You may request a letter from your doctor with specific instructions. Give this to the appropriate person at your child's daycare center, school or camp. You may also request an "emergency letter." Give this to the doctor treating your child during emergency room visits.

Do not be afraid to increase the medicine if you think your child needs a stress dose. A single increased dose will never cause harm, even if it isn't needed. Call your child's endocrinologist each time you give a stress dose. You should also feel free to call for advice at any time.

These are some of the times that a stress dose is needed:

During illness. Give your child a stress dose and then call the endocrinologist for any of the following:

- A fever of 101°F (38.3°C) by rectum or 100° (37.8°F) under the arm.
- Diarrhea (loose, watery stools when your child has a bowel movement).
- Repeat vomiting (throwing up more than once). **If this happens after giving your child the medicine by mouth, give your child a shot of the medicine. Call the endocrinologist and go the hospital right away.** This is an emergency and needs to be taken very seriously.

Let your endocrinologist know right away if your child is sick with fever, vomiting or diarrhea.

Major injury. If your child suffers a serious fall or broken bone, give your child a stress dose. Then, call your endocrinologist.

Surgery. If your child will have general anesthesia (medicine to make the child sleep during surgery), extra medicine is needed before, during and after the procedure. Make sure that the surgical staff knows this and consults with your endocrinologist.

Competitive sports. Sports that result in a lot of physical stress may require a stress dose during the competition. Again, ask your doctor what the right dose should be.

Regular doctor visits

Your child should see his or her endocrinologist every three to four months for blood tests, X-rays and an exam. As your child gets older, he or she will not have to go to the doctor as often.

Emergency hydrocortisone kit

Make sure that you always have a hydrocortisone kit at home for emergency use. This kit includes medicine you give with a syringe (a shot). It can be life saving. Your doctor will write a prescription and show you how to use it.

Your child should also wear a medical ID bracelet or necklace that reads “Adrenal Insufficiency—on hydrocortisone.” You may also wish to include the phone number for your endocrinologist.

Psychological counseling and support

With any lifelong condition, family counseling is helpful. Counseling should begin as soon as the diagnosis is made. It will also help to meet with other families who have a child with CAH.

Parents may feel a range of emotions when learning about their child's CAH, from shock and confusion to shame, anxiety, anger and sadness. Addressing these feelings will help the family accept the diagnosis and act in the child's best interest.

At times, people with CAH may struggle with shame and low self-esteem as a result of "being different." Parents and other adults will need to normalize the experience and provide emotional support and information. Contact with other people who have CAH is very helpful in turning shame into acceptance.

Other challenges may include body image concerns, insecurity with dating and sexuality, and concerns about possible infertility. All of this requires the support of parents, peers and health professionals. Specialized counseling may be useful throughout the person's life.

Surgery

In cases of some females with CAH, a question may arise about possible surgery to change the look of the child's genitals. Patients and parents should make this decision with the help of a psychologist and surgeon. Your doctor should offer you detailed medical information and all available options.

Untreated CAH

If your child does not take his or her medicine, it may lead to:

- Increased risk of a life-threatening adrenal crisis (in classical CAH).
- Entering puberty too early.
- Rapid growth in childhood and reduced height as an adult (because growth stops early).
- Unwanted body changes in females (excess body and facial hair, hair loss from the scalp, acne, enlarged clitoris, and irregular periods).

- Difficulty in having children (for both men and women).
- Tumors in the testicles for males and ovaries for females.
- Pituitary tumors in both males and females.
- Ovarian cysts in females.

CAH in children and young adults

Your child needs to be carefully watched for signs of early puberty and rapid growth. Early puberty is that which starts before the age of 8 in girls and 9 in boys. Signs include early pubic hair, early breast development in girls and enlargement of testicles in boys.

Rapid growth may affect your child's adult height. At first, the child will be taller than other children of the same age, but his or her final height will be reduced because growth stops too soon.

Making sure your child takes his or her medicine can prevent both early puberty and reduced height.

Frequently asked questions

At what age should I tell my child about his or her condition?

While there is no right or wrong age, it is important to always be honest and answer all of your child's questions. Telling the child is a gradual process. You will need to explain facts more than once.

As part of doctor visits and daily medicines, a child may ask, "Why do the doctors take blood from me every few months?" or "Why do I have to take this stuff?" These are excellent times to discuss CAH.

Who else should know about my child's CAH? How much should I tell others?

Parents need to weigh the pros and cons of telling others. Telling too little leads to a sense of secrecy, this can add to the parent's feelings of shame. Telling too much could result in the child being singled out, treated differently, and possibly teased and rejected by peers.

When your child is old enough, include the child in decisions about who should know outside of the family, including school medical staff and daycare providers.

Will CAH affect my child's height?

There are many factors that can affect a child's growth. Regular doctor visits are important because they help your doctor identify early warning signs of rapid growth, early puberty and advanced bone age. All of these may lead to a reduced height as an adult.

Your doctor will do regular blood tests to check hormone levels, take X-rays of the hand to check bone age, and chart your child's growth and physical development. This allows your doctor to prescribe the right treatment to help your child grow properly.

Is my child at an increased risk of infection because of being on steroids?

No. Some people who are on a high dose of steroids are at risk for infection because their immune systems are suppressed. However, your child's steroid dose will not harm his or her immune function. Your child is given just enough steroids to replace the hormones that the body should produce naturally.

Can my child receive live vaccinations?

Yes. Your child can receive all the normal childhood vaccines.

Glossary

Adrenal glands: Small glands located above each kidney that produce three important hormones: aldosterone, cortisol and androgen (DHEA, androstenedione, testosterone).

Aldosterone: Salt-saving hormone, made by the adrenal cortex. It acts on the kidney to help move salt from the urine back into the blood and to get rid of potassium.

Androgens: Male sex hormones that are made in the testes in males, ovaries in females and the adrenal glands in both males and females.

Bone age: X-ray of the hand that determines the age of bones in comparison to the chronological age.

Carrier: A person who has one out of two genes for an inheritable condition without being affected by the condition.

Clitoris: A small, sensitive organ located above the vaginal opening. Androgens make it grow larger.

Congenital adrenal hyperplasia (CAH): An inherited disorder of the adrenal glands resulting in insufficient cortisol production.

Cortisol: A steroid hormone made by the adrenal cortex. It maintains the body's energy supply and helps the body react to stress.

Estrogen: Female sex hormone, made by the ovaries in females and the adrenal glands in both males and females.

Gene: A unit of heredity located on a chromosome. It transmits a characteristic from parent to offspring.

Gland: Any organ or layer of cells that produces and secretes hormones.

Hormone: A chemical messenger made in a gland. Hormones are sent through the blood to target body organs and tissues, stimulating certain life processes and stopping others as needed by the body.

Ovaries: Female reproductive organs containing the eggs.

Pituitary gland: The "master gland," located in the brain. It regulates the thyroid, adrenal, sex and mammary (breast) glands.

Recessive genetic disorder: A disorder that does not show symptoms in a person unless two affected genes are inherited, one from each parent.

Salt wasters: Classical CAH patients who lack aldosterone (salt saving hormone). These patients have insufficient cortisol and high androgens. This disorder is life threatening if left untreated.

Simple virilizers: Classical CAH patients who have low cortisol and high androgens, but who are not salt wasters. This is not a life threatening condition.

Testes: Two egg-shaped male reproductive organs located in the scrotum. They produce sperm and the male hormone testosterone.

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