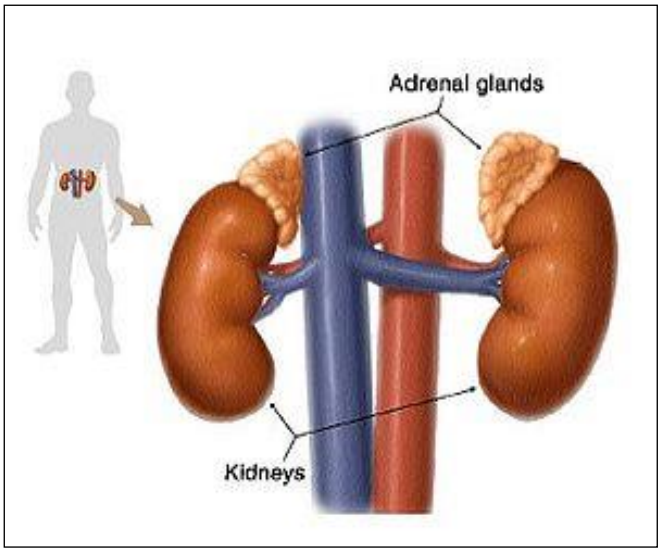
Congenital Adrenal Hyperplasia (CAH)

What is a hormone?

Hormones are chemical messengers. They are made in glands and travel round the body in the bloodstream. Hormones affect how other organs in the body work.

What are the adrenal glands?

The adrenal glands are two small structures on top of each kidney in the tummy.



The inner part of the adrenal gland (called the medulla) makes the stress hormones adrenaline and noradrenaline. These hormones are made in response to fright. This inner part of the adrenal gland works normally in congenital adrenal hyperplasia.

The outer part of the adrenal gland (called the cortex) makes three different hormones (sometimes called steroids):
Cortisol: controls the levels of energy and salt and sugar in the blood. Cortisol is also needed to help the body cope with the stress of infection or serious injury).
Aldosterone: stops the kidneys losing too much salt in the urine.
Androgens: sometimes referred to as male hormones but are made by the adrenal glands of both boys and girls.

What is Congenital Adrenal Hyperplasia (CAH)?

The term congenital adrenal hyperplasia (often shortened to CAH) describes the condition:
Congenital: means from birth
Adrenal: the glands affected
Hyperplasia: overgrowth or a thickening of the outer layer of the gland which happens as the adrenal cortex tries to make steroids but is unable to.

Why does it happen?

The adrenal glands make the steroid hormone from cholesterol. This is done by the action of several enzymes. Children with CAH are missing one of the enzymes needed for this so cannot make enough cortisol. 3 out of 4 children with CAH are also not able to make enough aldosterone. The body senses that there is not enough cortisol and tries to make the adrenal glands make more. Whilst trying to make cortisol the adrenal glands make too much of the androgen hormones.

Genes are like a set of instructions for our body that are inherited from our parents. Children with CAH inherit a gene from each parent that has a change in it. This means that the child cannot make the enzyme needed for their adrenal glands to make cortisol. This will be talked about more in clinic and you will also have an appointment with a specialist doctor called a Geneticist to find out more.

How does it affect a child?
This depends on how much of the enzyme the child is missing. Some children have no enzyme and so are not able to make any cortisol. These children will be more affected and will be found to have CAH at or soon after birth. Others are able to make some cortisol and so may not be found to have CAH until later in childhood.

The adrenal glands start to work before a baby is born. Babies who have very little of the enzyme will make far more androgens (male hormone) than usual. In baby girls this affects how their genital area develops. The clitoris gets bigger, the labia may be joined together and the openings for the vagina and urethra can be partly closed. These changes can mean that it is not possible to tell if the baby is a boy or girl when they are born.

Baby boys may have slightly darker skin than usual around their penis and scrotum but have no genital abnormality at birth.
As 3 out of 4 children with CAH cannot make enough aldosterone they will lose too much salt in their urine. This means the salt levels in the blood will become dangerously low within the first two or three weeks of life. The baby will not be gaining weight, is likely to be vomiting, dehydrated and unsettled. If this is not treated the baby will become critically ill and collapse. This is called an 'adrenal crisis' and can be fatal if not treated straightaway.

Girls and boys may who are less severely affected – so can make some of the enzymes may be diagnosed in early childhood when they develop pubic hair, body odour and are growing more quickly than expected.

Girls with a very mild form of CAH may not be diagnosed until late childhood or young adulthood due to excessive hair growth, irregular periods or difficulties getting pregnant.

Is it possible to make a diagnosis before a baby is born?
In a family with a child known to have CAH, genetic testing is possible to find out if an unborn baby is affected. Your child's consultant or your GP can refer you to a specialist genetics doctor who will be able to explain this to you and arrange for any tests that are needed.

Are there different types of CAH?
CAH is always caused by a child missing some or all of an enzyme needed to make cortisol.
Sometimes CAH is classified into different groups to help better understand the condition or decide on the best treatment options. The amount of the enzyme the child is able to make can be different even in children from the same family so the physical effects can be different too.

How is it treated?
CAH is treated by replacing the hormones that the adrenal glands are unable to make.

The tablet most commonly used in children to replace cortisol is known as hydrocortisone.

The tablet used to replace aldosterone is known as fludrocortisone. For babies affected by low aldosterone levels, extra salt (called sodium chloride) is usually also needed for about the first year to help them maintain their salt levels.

What about the high androgen levels?
Giving the child the hydrocortisone and fludrocortisone means that their body stops trying to make their adrenal glands work – so they should no longer be making too much androgen hormones.

Will my child develop normally?
The goal of treatment is normal growth and development as well as preventing health problems in later life. However, it can be hard to get the balance right between giving too little or too much steroids:

- Too much hydrocortisone causes weight gain and slowing of growth; too much fludrocortisone may cause high blood pressure
- Not enough hydrocortisone and fludrocortisone can cause poor weight gain, too much salt loss and high levels of androgens. This can lead to your child developing and growing too quickly. It can also mean an adrenal crisis is more likely to happen during fairly minor illnesses.

To start with you will need to bring your baby to clinic every few weeks. Once they are older they will still need to be seen in clinic at least twice a year so that you and the doctors can be sure the treatment is exactly right for them. At clinic appointments your child will be weighed and measured and examined by a doctor or nurse to look for any signs of early puberty. They will have their blood pressure checked and will need regular blood tests.

Can my child lead a normal life?
Yes. If your child has all the medicines as prescribed and is seen regularly in clinic they should be able to do everything their friends do. As an adult there will be some careers/jobs they will not be accepted for such as the armed forces. There are no restrictions on driving.

What happens if my child is unwell?
In a healthy person, the body is able to make more cortisol if needed, e.g. during illnesses, operations or other stressful situations. Children with CAH are unable to do this. They need to take extra hydrocortisone to help their body cope and stay well.

Your child will need double (or sometimes triple) their usual dose of hydrocortisone for childhood illnesses. If the child is seriously ill or has an accident or if the dose of hydrocortisone is vomited more than twice, the hydrocortisone must be given by injection. The child must then be taken to the children's ward or nearest emergency department for assessment and treatment.

All families with an affected child should have an injection of hydrocortisone available and know how to give it. Some children may have low blood sugars during severe illness; they will need extra carbohydrates or glucose at these times.

Your doctor or specialist nurse will give you more information about this.

Are there any long-term problems?
People with CAH usually have to take lifelong steroid medication. If treatment is well managed during childhood most people with CAH reach a normal height but are probably a little shorter than they would have been if they did not have CAH.
Keeping the right balance of steroids is important during adult life to avoid the complications of osteoporosis, obesity and hypertension.
Some adults have reduced fertility but there are an increasing number of options available to overcome this.

Suggested sites for further information:

www.livingwithcah.com

www.eurospe.org

www.apeg.org.au

www.caresfoundation.org

www.dsdfamilies.org

HNE Kids Health. Autosomal Recessive Inheritance Genetics
http://www.youtube.com/watch?v=Nv6qUsKYodA

This leaflet has been written by members of the BSPED & reviewed by the Clinical Committee. It is designed to give you some general information about your child's condition and treatment. Your child's doctor or specialist nurse will be able to answer any further questions you have about your child.

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