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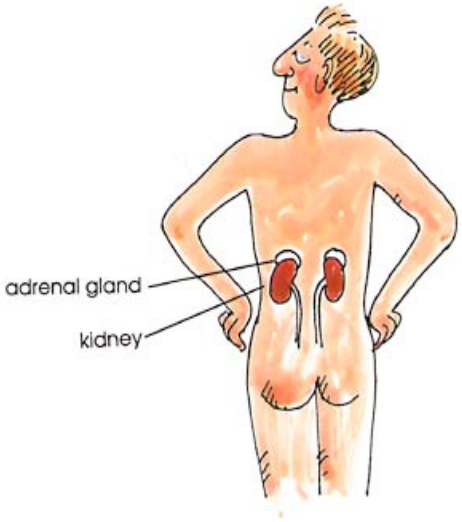
Congenital Adrenal Hyperplasia: Patient Education Material

Your child's been diagnosed to have CAH. To understand the condition, here is some information for you.

CAH is a fairly uncommon disorder, but one which is well understood and for which good treatment is readily available. People with CAH enjoy excellent health once treatment has been started, and live to a ripe old age.

The Nature of CAH

CAH is a congenital disorder (i.e. present at birth) involving the hormones of the adrenal glands. The word 'hyperplasia' means 'overgrown'. In CAH, the child is born with overgrown adrenal glands. The adrenal glands are a pair of fleshy triangular-shaped organs, each about the size of a walnut, which lie above the kidneys. The adrenal glands are controlled by the pituitary gland, a pea-sized gland at the base of the brain Hormones are the chemical messengers in the body.



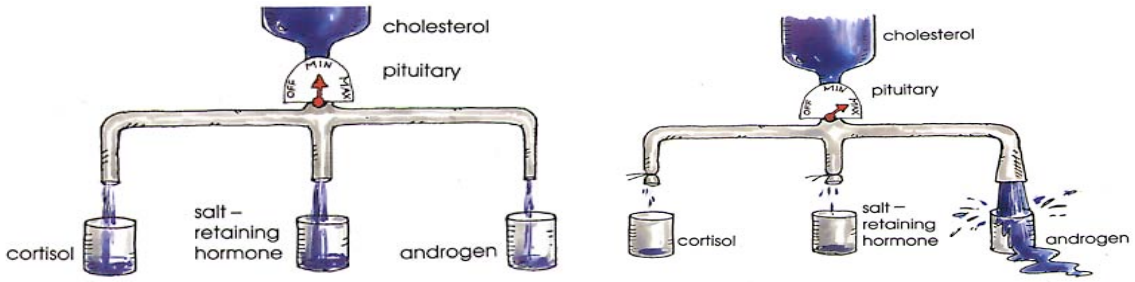
CAH - an enzyme deficiency- whether or not a person is born with the right number of enzymes in the body is determined by genetic or hereditary factors. In CAH, one of the adrenal enzymes is missing. This particular enzyme controls the production of both cortisol and the salt-retaining hormone. In CAH the excess androgens force the baby girls' genitalia to begin development in the male direction. Sometimes it is difficult to be sure about a CAH baby's true sex at the time of birth. Girls with CAH usually require surgery to restore the genital appearance to normal.

Adrenal Hormones

1. **Cortisol:** This is needed to protect the body from the generalised effects of illness or injury.
2. **Salt-retaining hormone**: This governs the amount of salt lost in urine via the kidneys.

3. **Androgen**: Hormones producing male effect. Both males and females have androgen. It is thought to aid growth in childhood.

In children with CAH, some of the complex chemical 'machinery' needed to make the essential hormones is defective, and the glands make the wrong hormones.



NORMAL CAH

Thus CAH results in three disturbances:
1. Lack of cortisol
2. Lack of salt-retaining hormone
3. Too much androgen

Treatment of CAH

CAH is a lifelong disorder for which no cure is available. We can, however, readily replace, in tablet form, the hormones that are missing – cortisol and the salt-retaining hormone. These medications will be needed every day for life, and need to be given even when the child is perfectly well. There is no need for a special diet in CAH. There are several medications available which can be used as substitutes for the natural hormone, cortisol. They are: hydrocortisone, cortisone, prednisolone and dexamethasone. They differ in potency (strength) and duration of action. If a salt-retaining hormone is needed, we use fludrocortisone. 'Florinef' or 'Flouricort' is its trade name. The exact dose of medication needed to treat CAH differs from one child to another, due to variation in body size, different rates of absorption from the bowel and other factors. As the child grows, the doses need to be increased as a matter of course. The reason for testing the blood and urine of CAH patients is to ensure that just enough medication is being given to control overproduction of androgen and no more. The test results are interpreted in conjunction with the growth of the child and other findings in physical examination.In the blood, we are interested in the levels of a particular chemical called 17-hydroxyprogesterone (17OHP) and electrolytes (Na and K).

Emergency condition: The adrenal crises- The body usually responds to sickness (fever, infection or other illness) and physical injury by releasing more cortisol. This helps to boost the blood sugar and blood pressure, aiding recovery. In CAH, the adrenal glands are unable to make more cortisol. Therefore the body is less protected, and the effects of illness or injury may be more severe. Adrenal crises can be readily prevented.

When to give extra hydrocortisone (Hisone)?

1. When your child feels ill, particularly if he or she has a high temperature (greater than 101^{o}F)
2. When your child requires a general anaesthetic for any reason.
3. When your child is vomiting, or has diarrhoea. Severe vomiting or diarrhoea in someone with CAH is often best treated in hospital.
4. Severe injury (e.g. broken arm or leg).

Extra hydrocortisone is not necessary with minor cuts and scratches, even if local anaesthetic is used.

How much extra hydrocortisone should be given?

About three times the usual daily dose should be given for 3-4 days. When hydrocortisone has to be given by injection, the doctor giving the injection will decide the dose.

If you are in doubt when to give extra hydrocortisone, it is always safer to give it, than not to give it.

Every person with CAH should carry a letter outlining the important medical problems that your child could have, and how these should be dealt with in an emergency.

If child is very sick/ hospitalization is needed: Inform the treating physician --------

Side-effects of hydrocortisone- The side-effects should not be seen in a CAH patient because in CAH the aim of treatment is simply to put back what is missing, bringing the levels up to normal. We can make sure that the dose is tailored exactly to the individual's needs by performing regular tests.

What are the risks in future pregnancies?

1. If a couple already has one or more children with CAH, and neither parent has CAH, the chance with each subsequent pregnancy that the child will have CAH, is one in four.
2. If a person with CAH marries one without CAH, the chance of their having a child with CAH is very low, about one in 100.
3. If two people with CAH have children, all of their children would have CAH.

CAH in an unborn baby can be diagnosed before birth. The newborn screening on dried blood spots collected 24-72 hours after birth is being used to detect infants with CAH before they become sick.

People affected with CAH can do anything they want to do, including marrying and having children. To maintain this state of good health, they do need to take medications, but this becomes accepted as part of life. There are many conditions much worse than CAH. CAH is a nuisance, but not a handicap.