What is congenital adrenal hyperplasia (CAH)?

The adrenal glands are located on top of the kidneys and produce several different hormones. Congenital adrenal hyperplasia (CAH) is a common inherited disorder in which the adrenal gland cannot make enough of a hormone, called cortisol, or a salt-preserving hormone, called aldosterone. The adrenal glands are located on top of the kidneys and produce several different hormones. Aldosterone helps the body hold onto sodium and release excessive amounts of potassium. In addition to cortisol and aldosterone, the outer portion of the adrenal gland (adrenal cortex) makes male-like hormones, called androgens. Some patients with CAH also cannot make enough aldosterone. The male-like hormones are responsible for the normal development of adult-type body odor, underarm hair, pubic hair, and adolescent acne. The inner portion of the adrenal gland (adrenal medulla) makes epinephrine, often called adrenaline.

The symptoms of the severe classic type of CAH are apparent at birth in girls in the form of a male-like genital appearance known as genital ambiguity; boys, however, often may not have any physical signs. Screening performed through newborn screening programs shortly after birth helps to identify most of these newborns. Other individuals do not develop symptoms until early childhood, the teenage years, or even adulthood. This form of CAH is called non-classic or late onset.

Cortisol is important for normal health and well-being. Symptoms associated with low cortisol levels are often nonspecific, but can include nausea, repeated vomiting, poor appetite, weight loss, and fatigue. Symptoms associated with low aldosterone levels may include low blood pressure, dehydration, low blood sodium (salt) levels, and high blood potassium levels.

The adrenal gland cannot make enough cortisol or aldosterone in classic CAH; instead, it makes large amounts of extra male-like androgen hormones. High levels of androgen hormones can alter the appearance of the external genitals of newborn girls to look more like those of a newborn boy (a condition known as virilization). In such cases, newborn girls may have an enlarged clitoris, labia (the lips around the opening of the vagina) that are fused together, or genitals that look very similar to those of a boy, except that the testicles are missing. Baby boys with CAH usually have normal male external genitals; their testicles are located in their scrotum.

Young children with non-classic CAH may develop pubic hair at ages younger than normal or may develop enlargement of the clitoris or penis. Teenaged and adult women with non-classic CAH may present for medical care because of irregular menstrual periods or excessive body hair (hirsutism).

How common is CAH?

The most common type of CAH is caused by 21-hydroxylase deficiency due to abnormalities in the 21-hydroxylase (CYP21A2) gene. This gene tells our bodies how to make the 21-hydroxylase protein. When the 21-hydroxylase protein is absent or does not work normally, the adrenal gland cannot make enough cortisol. For some patients, the adrenal gland cannot make enough cortisol or aldosterone.

Congenital adrenal hyperplasia is a hormone deficiency disorder. The main treatment is hormone replacement with cortisol. This hormone is called hydrocortisone, but it is also available under various trade names, such as Cortef. This medication is usually given by mouth 3 times each day. For patients with aldosterone deficiency, fludrocortisone is usually also necessary, and for some of these newborns, additional salt solutions by mouth may be needed. It is important that a pharmacist prepare all medications, including any salt solutions, for patients with CAH.

Your child will require follow-up visits and careful monitoring by a pediatric endocrinologist to make sure that your child is growing and de-
veloping normally and is given the most appropriate hormone replace-
ment treatment. With appropriate hormone therapy, typical growth,
development, and fertility are highly possible.

Individuals with classic CAH must take extra cortisol dosages when they
are sick to maintain health. Your child’s physician will teach you when
and how to increase the cortisol dose. Your child’s physician will also
teach you how to administer cortisol by an intramuscular injection (eg,
Solu-Cortef, Hydrocortisone sodium succinate) when your child is unable
to take the medication by mouth. Individuals with CAH should wear med-
ical alert identification badges. Although many patients with non-classic
CAH may not need to take daily cortisol replacement dosages, they may
need to take cortisol replacement dosages during a major illness, such as
high fevers or during periods of significant physical stress.

Can CAH be prevented?
CAH cannot be prevented. In the past, an experimental medication was
prescribed for pregnant women who were at risk for having a newborn girl
with CAH to prevent the overgrowth of the clitoris and atypical external geni-
tal appearance. This experimental medication does not cure CAH. In addition,
current information indicates that this treatment may have negative effects in
later life and is no longer recommended.

Families concerned about their risk of having another child with CAH
should discuss this concern with their child’s endocrinologist in consul-
tation with a genetic counselor.

Pediatric Endocrine Society/American Academy of Pediatrics
Section on Endocrinology Patient Education Committee

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN®