Fact Sheet: Classical Congenital Adrenal Hyperplasia (CAH): Adolescents and Adults

Congenital Adrenal Hyperplasia (CAH) is a family of inherited disorders affecting the adrenal glands. 21-hydroxylase deficiency is the most common form of CAH. The disorder is divided into three distinct forms: salt-wasting CAH, simple-virilizing CAH, and the mild form known as non-classical CAH.

Classical CAH is subdivided into the salt-wasting form and simple-virilizing form. In both forms the deficiency of the 21-hydroxylase enzyme is severe, which leads to inadequate levels of cortisol, a hormone that governs the body's energy supply, blood sugar, and reaction to stress. The deficiency also causes an elevation of male-like hormones, called androgens. Because the androgens are already high in the womb, baby girls with classical CAH are born with abnormalities of the genitalia and many have reconstructive surgery in childhood and later on in life.

In childhood high androgens can cause rapid growth, which can lead to a child being taller than his peers early on, yet growth plates can close early, leading to short stature in adulthood. High androgens can cause acne, male-pattern baldness and infertility in both men and women. Women may develop excess body hair. Women must also be monitored for development of polycystic ovarian syndrome, a disorder where the ovaries produce high androgen levels. Men must be monitored for development of testicular adrenal rests, which are benign masses in the testes that can lead to low libido and infertility.

People with salt-wasting CAH are also deficient in aldosterone, a hormone responsible for salt, potassium, and water balance. People with simple-virilizing CAH do not have aldosterone deficiency.

Having CAH can certainly affect a person's quality of life; however, treatment of the disorder has been well-established, and with compliance with treatment many of the above symptoms can be prevented. Daily dexamethasone and/or hydrocortisone are given in both salt-wasting and simple-virilizing forms. These medications replace cortisol that is deficient in patients with classical CAH, ensuring adequate energy and steroid metabolism. In addition, these medications are aimed at decreasing the level of androgens. When you become ill, have a fever, are injured or are undergoing surgery, higher “stress” doses of hydrocortisone are given. Patients with salt-wasting CAH are also prescribed fludrocortisone (Florinef), a medication that behaves similarly to aldosterone and helps the body maintain adequate salt and potassium levels. Patients with salt-wasting CAH frequently crave salty foods and should not limit salt in their diet, unless instructed by your endocrinologist. Medication doses are adjusted frequently based on your hormone levels. In women adjuvant treatments may be utilized to improve signs and symptoms of elevated androgens.
Our team is here to help you every step of the way.

- Our team of physicians will see you frequently in the beginning; however, once your adrenal hormones have been well-controlled and symptoms have improved, visits can take place every 6 months. We are available 24 hours a day, 7 days a week should an emergency arise. Our number is 212-746-3975.

- Our surgical team is one of the leading teams in the world specializing in genital reconstructive surgery. Men with testicular adrenal rests should have an evaluation by a urologist as well. To make an appointment with a surgeon, please call 212-746-5337.

- The Cornell Institute of Reproductive Medicine is an internationally recognized reproductive center where a referral will be made when necessary.

- Our psychologist, with years of experience with patients and families living with a chronic illness, can provide ongoing psychological support.

- Our nutritionist will work with you on a proper diet should weight control be an issue.