Cortisol hormone levels. When cortisol levels are low, the body's master gland (called the pituitary) produces more adrenal stimulating hormone (called ACTH) which stimulates the adrenal glands to produce more cortisol. This leads to hyper trophy and enlargement of the adrenal glands as they try to produce more cortisol but are not able to due to the enzyme deficiency. The excess 17-hydroxyprogestosterone is metabolized to unwanted excess androgens in individuals with CAH and can be measured as androstenedione, testosterone, and other markers. ACTH stimulation testing can be done clinically to look at adrenal hormone levels, and this test can be helpful in establishing the diagnosis of CAH.

The gene for the 21-hydroxylase enzyme can also be evaluated by DNA analysis which determines whether the gene is present or absent, or if the DNA make-up is changed (mutated) in the patient. By obtaining a blood sample, mutations can be identified in the affected individual. Genetic testing can also be done in other family members to see if they carry the mutations. Carrier detection serves the important function of alerting the person to the possibility of having a child affected with CAH. Genetic counseling can be done in those who are carriers for the CAH gene.

Newborn Screening for CAH
Newborn screening for CAH has been mandated by law in the United States since 2008. It is performed by measuring the 17-hydroxyprogesterone level in a tiny blood spot obtained on a filter paper by heel prick of newborns usually at 24 to 48 hours of life (the time of the collection varies in each state). The screening test is performed by the laboratory of Department of Public Health in most state’s regional programs. Check with your regional Department of Public Health on newborn screening for CAH for current status.

Prenatal Diagnosis
When there is a family member with CAH, it is possible to diagnosis a child before birth through tests performed during pregnancy. However, prenatal therapy is considered experimental as the risks and benefits of this procedure to the mother and to the affected child are unclear. Prenatal therapy should only be done at medical centers under approved research protocols.
Treatment of CAH in children, adolescents, and adults

The aim of treatment is to provide the body with the ability to maintain a normal energy level, balance of salt and water, normal growth, sexual maturation at an appropriate age, and fertility later in life. This is accomplished by replacing the inadequately produced cortisol hormone by a synthetic glucocorticoid. Hydrocortisone is the preferred glucocorticoid in growing children. Other glucocorticoids that are also commonly used are prednisone or prednisolone. Dexamethasone is a very long-acting glucocorticoid that is also sometimes used, but some physicians worry about the strong potency and long-term effects on linear growth. Steroid dosing needs to be individualized and doses vary between individuals with CAH. In salt wasters, a synthetic salt retaining hormone called fluocortisone helps to retain sodium and water in the body. In infancy, salt replacement is also needed. The glucocorticoid medication is dosed based on body surface area and then adjusted based on hormone levels (e.g. 17-hydroxyprogesterone, androstenedione, testosterone) in the blood. The fluocortisone medication is dosed based on blood tests (plasma renin activity level and electrolytes) as well as blood pressure measurements. Too much medication can lead to various problems, such as poor linear growth, short stature, weight gain, reduced bone density, and elevated blood pressure. Too little medication can also lead to problems, such as sodium and potassium imbalance, improper growth, advanced bone age, early pubic hair, acne, and fertility problems. Therefore, the care of children with CAH is complex. Treatment of CAH is ongoing, involving periodic medical evaluations, monitoring for medication dose adjustments, and checking compliance.

The virilized female genitalia may require corrective surgery as an infant, and if needed, again later in life. Talking to an experienced Pediatric Urologist about potential surgeries and the timing of the procedures is important.

Special care needs for CAH

Without cortisol, the body cannot respond to stress. A child affected with CAH can go into shock from infection, injury, or surgery. Extra doses of glucocorticoid are important at these times of stress. Therefore, during illness, the oral dose of synthetic glucocorticoid is tripled. However, if the child is not able to take the medication by mouth due to vomiting or cannot absorb the medication due to severe diarrhea, the parents should give the child an emergency injection of hydrocortisone into a muscle. They should then notify their child’s doctor and potentially take child to their local emergency department for evaluation. When the child is ill and receiving stress oral steroids, they should drink sugar containing liquids (and not just water) in order to avoid hypoglycemia. It is recommended to carry a special care instruction prepared by your child’s endocrinologist to the emergency department so that they are aware of the importance of your child receiving appropriate care with fluids and stress doses of hydrocortisone. CAH is a relatively rare condition and all doctors may not be aware of it.

If at any point a child with CAH requires surgery, it is imperative that the child’s endocrinologist be notified. For the stress of surgery, a child with CAH will require special doses of hydrocortisone during and after the surgery. The endocrinologist will be able to inform the other doctors of the necessary precautions. It is also mandatory to have your child wear a Medical Alert ID bracelet stating that your child has “adrenal insufficiency” or that they are “cortisol dependent”. In an emergency, this will alert health care personnel to the importance of your child receiving appropriate stress doses of glucocorticoids.

Treatment of CAH is life-long. Periodic medical check-ups, laboratory monitoring, and compliance with medications allows for a full and otherwise normal healthy life. Recommended reading (free full text on-line):


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Congenital Adrenal Hyperplasia

The MAGIC Foundation is a national nonprofit organization created to provide support services for the families of children afflicted with a wide variety of chronic and/or critical disorders, syndromes and diseases that affect a child’s growth. Some of the diagnoses are quite common while others are very rare.

Recommended reading (free full text on-line):


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