Congenital Adrenal Hyperplasia

What is Congenital Adrenal Hyperplasia?

Congenital Adrenal Hyperplasia (CAH) is an inherited disorder of the adrenal glands. The adrenal glands located at the top of the kidneys, produce three types of hormones called cortisol, aldosterone and androgens. The life essential hormones are cortisol and aldosterone. Cortisol is an energy and blood sugar regulating hormone. Aldosterone is a salt and water-regulating hormone. Androgens are "masculine hormones" that cause pubic hair growth and masculine changes in puberty, called viralization. Normally, there is an enzyme (a protein in the body that causes a chemical change) that is necessary to make cortisol and aldosterone. In CAH the enzyme is either missing, partially inactive, or totally inactive which leads to a "roadblock" in the "hormone production highway". If the enzyme is missing or severely inactive, both cortisol and aldosterone will not be produced. If the enzyme is partially inactive, the cortisol produced will be reduced. In addition, the other hormones which are in the same or different pathway "highway" to make cortisol and aldosterone are also affected and may be abnormally low, or abnormally high. ACTH, produced from the pituitary gland, which is located at the base of the brain, stimulates the adrenals to make cortisol. When the cortisol level is low, the pituitary will send more ACTH to the adrenals in an attempt to make more cortisol. In CAH, this results in the overproduction or "backup" of the other adrenal hormones just before the "roadblock" enzyme occurs- .

CAH can be virilizing (masculinizing) or feminizing, depending on the inactive enzyme. The virilizing type is caused by the overproduction of androgens (masculine hormones). The feminizing type is caused by lack of necessary male hormones in the males that result in a more female appearance of the genitalia. The missing enzyme also determines if the CAH is a salt wasting or non salt wasting type. Salt wasting applies to children who lose salt from the kidney due to lack of aldosterone. Non-salt wasting type applies to children who produce enough aldosterone, but not enough cortisol. Usually people with non-salt wasting CAH have a milder degree of CAH.

What causes CAH?

CAH is a congenitally inherited disorder. This means that your child was born with the disorder. The "pattern" for the baby is contained in the chromosomes (genes) inherited from the parents. The chromosome for the missing or inactive enzyme was passed on to the child from both parents. In general, parents of children with CAH are healthy, but each parent carried the partial trait for CAH.

There are several forms of CAH, depending on which enzyme is deficient. Ask your physician which enzyme is missing or inactive in your child's type of CAH. The treatment and symptoms will vary according to the type of CAH. The most common type of enzyme deficiency that causes CAH is 21 hydroxylase (21OH deficiency). Other adrenal enzyme deficiencies that less frequently cause CAH are steroidogenic acute regulatory (StAR) protein, 3ß-hydrozysteroid dehydrogenase, (3ßHSD), 11ß-hydroxylase (11ß-OH, and 17ą –hydr4ozylase (17ąOH) deficiencies.

What are the possible effects of CAH?

There are several effects that may be seen in CAH. The primary problem in CAH is low cortisol. Cortisol is the hormone that helps your body react to stress. It also plays an important role in

regulating blood sugar and the body's energy supply. If your child is lacking the salt retaining hormone, she/he may also have difficulty maintaining body salt and water balance. Low cortisol and aldosterone productions are the reason that newborns and children not receiving treatment get very sick. This is called an adrenal crisis and is life threatening. Signs of adrenal crisis in infants include poor feeding, vomiting and diarrhea, sleepiness, weak cry, dehydration, low blood pressure, weight loss and poor growth. In severe cases, the androgens (male hormones) can affect the baby's genitalia before birth. Baby girls may be born with genitalia that may appear male-like, while the baby boy's genitalia will appear normal. In otherforms of CAH, baby boys may be born with genitalia, which appears female-like. Some children with CAH may have high blood pressure.

Some children with a milder form of CAH may not show any symptoms until they are older. This form is sometimes called late onset CAH. The excess androgens may cause rapid growth, advanced bone age and early development of acne and body hair. Even though these children are taller than their same-aged friends, during childhood, they may become a short adult due to the rapid maturation of the bone. The teenager or adult with CAH may have excess body hair, cystic acne, and in girls, menstrual irregularities and fertility difficulty.

How is CAH diagnosed?

CAH is diagnosed by a blood sample test from your child. Cortisol and the other adrenal hormone levels will be measured. For example, if your child has 210H deficiency CAH, the blood test would most likely show a low cortisol and a high 170H Progesterone and androgen level. Your physician may also request a blood test before and after adrenal stimulation (a blood test that requires multiple samples). CAH is a genetic disorder in which the gene that makes the enzyme is defective (parts of the gene may be missing or rearranged). The genetic cause of CAH can be determined by doing a blood test that looks directly at the structure of the DNA gene. Other family members may find out if they are carriers of CAH by having similar blood tests.

Your physician may order additional tests such as bone age X-rays or ultrasounds of the abdomen and pelvis, to visualize the adrenal glands or the structure of the internal genitalia.

If you have a child with CAH, and have another pregnancy, tests may be done to determine the baby's sex and whether the unborn has CAH. If the fetus is a female and has CAH, prenatal medication may be given to prevent the development of male-like external genitalia.

What is the treatment for CAH?

The female baby may require surgery to repair and complete the process of sexual anatomy development. The timing of genital surgery is a decision that parents will make with the multidisciplinary team.

HORMONE REPLACEMENT

Cortisol hormone replacement therapy will be prescribed in the form of hydrocortisone. If your child has the salt-wasting type of CAH the aldosterone will be replace by florinef. When your child is sick he/she will need a stress dose of cortisol replacement. If your child vomits and/or is unable to take the cortisol by mouth an intramuscular injection (shot) of cortisol will be necessary. Your physician will give you prescriptions for all medications. It is essential to follow the directions.

Sick Days

Any illness or other physical stress may affect your child's health. Generally, physical stress is defined as fever higher than38 degrees Celsius, strep throat, ear infection, pneumonia, vomiting, muscle strain or sprain, broken nose, or a serious accident that requires immediate medical

attention or hospitalization. During time of illness or stress the body needs larger amounts of cortisol to heal. People with CAH do not make enough cortisol to help the body to recover from illness or injury. They will need to be given extra cortisol (stress does). When ill encourage your child to drink extra fluids (8-12 ounces every hour while awake) to help prevent dehydration. If the illness includes vomiting and/or diarrhea your child will need extra cortisol, but if given by mouth it will not be absorbed. Therefore, he/she may need to be given the cortisol by injection (stress IM dose). It is a good idea to ask about sick day management during your child's regular doctor visits so that you will be prepared. If there is any question about the need for additional treatment, check with your physician or nurse.

Emergencies

If your child "looks bad" (pale, sweaty or breathing fast), feels weak and/or is unable to respond normally, loses consciousness, or has a serious injury, such as a broken bone, **call 911 and give the cortisol injection as previously instructed by your physician or nurse. This is a life-threatening emergency and your child needs an injection of rapid-acting cortisol immediately!**

Safety

Ideally, the parent should learn to give a cortisol injection to prevent a life-threatening emergency. To be prepared for an emergency, it is essential to obtain a vial of injectable cortisol and carry it with your child at all times. It is also recommended that your child wear personal identification, such as a Medic-Alert bracelet or necklace that identifies that he/she has CAH. This will ensure that emergency medical personnel will know how to quickly treat your child in your absence.

Other Issues

Children with CAH are capable of participating fully in school activities, sports and social events. If properly treated, girls with CAH can have regular menstrual cycles and bear children. Pregnancies are monitored closely by an endocrinologist to assure the best medication dosage for the mother. With proper treatment, these children are expected to have a normal life span.

When should the physician see my child?

Once the condition is stable, the endocrinologist normally sees children with CAH every 4-6 months. Additional office visits may be necessary depending on the child's age and response to treatment. Your child will have blood tests to check the hormone levels and the need to adjust the medication. Height, weight, bone maturation, and sexual development will be followed. If your child's genitalia is affected he/she may need to follow-up with the genitourinary physician. Remember to continue office visits to your child's primary care physician for routine medical care.

If you have additional questions, your contact is _____

Phone # _____