Overview

Congenital adrenal hyperplasia (CAH) is a treatable disorder that occurs when the adrenal glands do not function properly. The result is that normal amounts of important hormones are not produced. If left untreated, CAH will cause problems with growth and development and can cause life-threatening illness. There are several forms of CAH, but one specific enzyme problem accounts for over 90 percent of diagnosed cases. This form is called 21-hydroxylase enzyme deficiency. Several other enzyme deficiencies involved in the production of adrenal hormones can also cause CAH. The only form of CAH detected by Kansas newborn screening is 21-hydroxylase deficiency.

What is congenital adrenal hyperplasia (CAH)?

Congenital adrenal hyperplasia (CAH) is an inherited condition which causes an enzyme deficiency (most commonly 21-hydroxylase) resulting in the inability of the adrenal glands to make hormones necessary to maintain life. The adrenal glands produce different hormones necessary to help the body to function normally. The three main hormones involved in CAH are:

- Cortisol - affects the body’s energy supply, blood sugar, blood pressure and control of the body’s reaction to stress.
- Aldosterone - maintains the salt and water balance in the body.
- Androgens - male sex hormones that affect body growth and appearance of external genitalia.

In a person with CAH, the adrenal glands are unable to properly regulate the hormones they produce. The result is not enough cortisol and aldosterone, and too much androgens. CAH can affect both boys and girls.

Why is newborn screening done for CAH?

Newborn screening is done for CAH so that babies with this condition can be diagnosed quickly. Some forms of CAH are very severe. They cause adrenal crisis resulting in low blood sugar and blood salt abnormalities that can be life threatening in the newborn due to salt wasting. Infants with severe CAH who are not diagnosed and treated early are particularly susceptible to sudden death in the first few weeks of life.

Does a presumptive positive result from the Kansas Newborn Screening Lab mean that my baby has CAH?

No, not necessarily. Newborn screening tests the baby's blood level of 17-hydroxy progesterone. That level was high in your baby. Prompt follow up is important. In addition to CAH, a few other medical problems can cause your baby’s abnormal test. Additional tests will need to be done to determine if your baby has CAH or not.

How common is CAH?

Classical CAH occurs in approximately one of every 15,000 – 25,000 births, while the non-classical form is more common.
How is CAH inherited?

CAH is inherited in an autosomal recessive pattern. Parents of a child diagnosed with CAH are unaffected. These individuals are carriers of the condition. Each pregnancy between carrier parents has a 25% chance of producing a child affected with CAH, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

What are the signs and symptoms of CAH?

The severe forms of the disorder (called "classical" CAH) can result in ambiguous genitalia (outer genitals do not have the typical appearance of either a boy or a girl) in a newborn girl. Infants with symptoms of impending adrenal crisis include vomiting, dehydration, weight loss, poor feeding, electrolyte imbalance and extreme tiredness.

How is CAH diagnosed?

Diagnostic tests include serum 17-OH progesterone (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced). Additional tests may be recommended by a specialist.

Is there a cure for CAH?

Right now, there is no cure for CAH, but there is treatment. Some people with mild CAH may not need to take medicine all the time. They may only need to take cortisol when they are sick. Most people with the classical form of CAH need to take cortisol every day.

How is CAH treated?

Newborns are placed on steroid hormones as soon as possible. These hormones must be taken for a person’s entire lifetime. Children with the salt-wasting form of CAH may also require salt supplements. Reconstructive plastic surgery on external genitalia may be needed for some newborn baby girls with CAH. Regular visits to a doctor specializing in diseases that affect hormones (endocrinologist) are needed to monitor the amount of medicine required by a child. Your pediatric endocrinologist and pediatrician will ensure your child is prescribed the proper amount of medicine for his or her own unique needs and growth.

Where can I get additional information?

CARES Foundation: www.caresfoundation.org

The ABC’S of Congenital Adrenal Hyperplasia: www.dshs.state.tx.us/newborn/cahbroch.shtml

GeneTests: www.genetests.org

MAGIC Foundation for Children's Growth: http://www.magicfoundation.org
Phone: 708-383-0808 or 800-362-4423   Fax: 708-383-0899