

KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

Kentucky Newborn Screening Program, 275 East Main Street, Frankfort, KY 40621, Phone (502) 564-3756, Fax (502) 564-1510

Congenital Adrenal Hyperplasia (CAH)

CAUSE In CAH, the child is born with overgrown adrenal glands, which are unable to make enough cortisol. Cortisol is essential for life as it is needed to maintain an adequate energy supply and blood sugar level. Cortisol is also called a “stress hormone” because our adrenal glands make more of it when we are under physical or emotional stress to protect our bodies from illness or injury. Another important hormone made by the adrenal gland is called aldosterone, a salt retaining hormone. Aldosterone is used by the kidney to help the body maintain normal levels of sodium and potassium, which are necessary for all cells to work normally. Without adequate levels of these minerals in the body, too much salt and water are lost in the urine, leading to salt deficiency and dehydration. Children with CAH also produce too much androgen (male hormone). The overproduction of male hormone in female infants will cause the external genitals to take on a male appearance, although the internal parts may be quite normal. For this reason, females are more likely to be diagnosed earlier than boys. Boys are likely to be diagnosed as a result of entering puberty at an extremely early age (some as young as 2-3 years old) and appearing to be very tall for their age.

IF NOT TREATED Untreated CAH can result in serious illness due to salt-wasting and, in some cases, death. It may be difficult to tell if the CAH child’s genitals are male or female. Without treatment, the clitoris in the female infant will continue to enlarge into male-like penile structure. Untreated females do not have normal periods and are unable to have children as an adult. The testicles of a boy with CAH cannot function well and will not make sperm normally. High levels of androgens cause rapid early physical growth that will also stop prematurely, resulting in the child becoming a very short adult.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist. CAH cannot be “outgrown” and treatment will be needed throughout life.

- CAH is treated with hormone-replacement medications. The metabolic specialist will determine the proper amount your child should take each day. These medications will require a prescription.
- Reconstructive surgery may be required for girls with masculine external genitalia and can be performed as early as one to three months of age.
- Contact your child’s doctor immediately at the start of any illness. Children with CAH may need to be treated at a hospital to prevent serious health problems.
- Blood tests will need to be performed frequently to monitor hormone levels and adjust medications as necessary.

IF TREATED

With regular medication, your child with CAH can lead a normal life. There are no known mental delays associated with CAH. Life expectancy is normal. Women with CAH that take their medications as directed have no difficulties becoming pregnant and carrying a baby to term. Men who take their medications appropriately also have normal fertility.



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