



State of CT Genetics Newborn Screening Program Family Fact Sheet

CONGENITAL ADRENAL HYPERPLASIA (CAH)

Hormones are an important part of our body. Babies who have the disorder called congenital adrenal hyperplasia produce low levels of one or two adrenal steroid hormones, cortisol and aldosterone. This causes the increased production of other hormones called androgens or male steroid hormones. Babies with congenital adrenal hyperplasia (CAH), have very high levels of androgen hormones in their body. High levels of androgen hormones can cause variable degrees of masculinization of female genitalia. Deficiency of aldosterone can cause loss of sodium (salt) and dehydration. In the most severe type, infants can become critically ill with vomiting and dehydration.

Connecticut began screening for CAH in 1997. In Connecticut, about 1 in every 32,930 newborn babies has CAH.

How does a baby get CAH?

CAH is an inherited disorder. Both the mother and the father have at least one abnormal gene that results in too little cortisol/aldosterone and too much androgen. A baby has to receive this abnormal gene from both the mother and the father to have this disorder.

How is a baby tested for CAH?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State laboratory to screen for CAH. Abnormal screening results are reported to the Department of Public Health, Genetic Newborn Screening Tracking Unit nurse consultant. If the results are borderline, the DPH nurse consultant will notify the baby's primary care provider of the need to obtain a second specimen. If the results are presumed positive, the DPH nurse consultant will notify the baby's primary care provider of the need for a prompt referral to one of the Endocrinology Regional Treatment Centers at Yale or Connecticut Children's Medical Center for confirmation testing and follow-up treatment if necessary.

What is the treatment for CAH?

If this diagnosis is confirmed, the Regional Treatment Center specialists will prescribe treatment for CAH, which includes supplements of the missing hormones. Early diagnosis of CAH leads to early treatment, which prevents life threatening illness and allows for normal growth and development.



Keeping Connecticut Healthy

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