

Congenital Adrenal Hyperplasia Fact Sheet for Providers

What is Congenital Adrenal Hyperplasia?

Congenital Adrenal Hyperplasia (CAH) is an inherited disorder that affects the adrenal glands and a patient's hormone levels. Specifically, a patient cannot produce cortisol, may not produce enough aldosterone and may produce too many androgens.

If untreated this condition can lead to serious illness and affect the child's development. Children with CAH are at risk of adrenal crisis, which can result in shock and death.

What are the symptoms of Congenital Adrenal Hyperplasia?

Symptoms of CAH may include:

- Lethargy
- Vomiting
- Poor feeding
- Ambiguous genitalia
- Failure to gain back birth weight by 10-14 days

How is Congenital Adrenal Hyperplasia identified and diagnosed?

All infants with an abnormal CAH newborn screening will require a second screening within 24 hours. If that screening also comes back positive for CAH, or if the child's electrolyte levels are abnormal, they will require a confirmation test measuring serum levels of 17-hydroxyprogesterone (17-OHP).

How is Congenital Adrenal Hyperplasia treated?

The main treatment for CAH is to replace the hormones that are missing. To replace cortisol, a medication called hydrocortisone is administered on a daily basis. Higher doses of hydrocortisone will be required if the child is sick, injured, or requires sedation/surgery. For patients with aldosterone deficiency, an additional medication called fludrocortisone is also usually necessary. Some newborns will also require additional salt supplementation.

Hormone therapy is life-long. For children with abnormal genitalia, surgery may be considered in consultation with specialty surgeons.

Where do I go for more information?

Use your smart phone's camera to scan the QR codes below.



Endocrine Society: https://www.endocrine.org/-/media/endocrine/files/patient-engagement/patient-guides/patient_guide_the_truth_about_congenital_adrenal_hyperplasia.pdf



Pediatric Endocrine Society: <https://www.endocrine.org/patient-engagement/endocrine-library/congenital-adrenal-hyperplasia>

How do I handle an abnormal screen for Congenital Hypothyroidism?

Early treatment is critical to make sure the baby does not go into adrenal shock. In the case of an abnormal screening, IMMEDIATELY take the following steps WITHIN 24 HOURS:

- Examine the newborn for symptoms of CAH, including virilization or life-threatening dehydration
- Check electrolyte levels and continue to monitor
- Perform a repeat newborn screen within 24 hours and send to the NC newborn screening laboratory by overnight delivery (or Monday delivery if weekend)
 - Be sure to mark as a REPEAT newborn screen
 - Do not expose the sample to heat or sunlight or place sample in a plastic bag for mailing
 - If mailing by overnight delivery send to:
 - NC State Laboratory of Public
Health Newborn Screening
Section
4312 District Drive
Raleigh, NC 27607
 - UPS and FedEx guarantee overnight delivery, US Mail does not.
- Continue to monitor the baby while the repeat screen is pending. Check electrolytes every few days.
- IF ELECTROLYTES OR REPEAT NEWBORN SCREEN ARE ABNORMAL, OR IF CLINICALLY INDICATED, collect a serum sample for 17-OH progesterone and send to one of the following:

Esoterix, Endocrine Sciences-
LabCorp 4301 Lost Hills Road
Calabasas Hills, CA 91301
(800) 444-9111
www.esoterix.com

OR

Mayo Clinic
Laboratories
3050 Superior Drive
NW Rochester, MN
55905 (800) 533-1710

- Consult with pediatric endocrinology
- If the baby is confirmed to have CAH, please consider referring for genetic counseling



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