



Congenital Adrenal Hyperplasia

What is newborn screening?

Newborn screening is a blood test to check for conditions that might be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel soon after birth. This blood sample is required for all newborn babies.



Newborn screening is not the same as diagnostic testing. A diagnostic test can tell with more certainty whether or not a child has a condition. On the other hand, a screening test simply indicates that a child

may have a condition. The purpose of a screening test is to find babies that should have diagnostic testing. When a child with an out-of-range newborn screening result has a follow-up test result within the normal range, it is sometimes called a "false positive."

What if my baby needs more testing?

If you are told that your baby needs follow-up testing, it does not necessarily mean that your baby is at risk. An out of range result may occur because:

- The sample was too small
- The sample was collected too early
- The sample was collected too close to a feeding
- The baby was born too early or had a low birth weight

Some babies who have follow up testing for congenital adrenal hyperplasia are healthy, and will not be diagnosed. However, out of range screening results CAN indicate a disorder, so it is important to follow your doctor's advice & get your baby tested quickly so that final results can be confirmed.

KEY POINTS:

- **You have just heard that your baby may have congenital adrenal hyperplasia. Please understand that the newborn screening is just that---a screening test. Further testing is required to confirm or rule out the diagnosis.**
- **Many babies who have out of range newborn screens are healthy, and will not be diagnosed.**
- **If treated early, children with congenital adrenal hyperplasia can have healthy growth and development.**

Connecticut Department of Public Health

Connecticut Newborn Screening Program • 860.920.6628

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Adapted, with permission, from the Minnesota Department of Public Health

What is congenital adrenal hyperplasia?

Congenital adrenal hyperplasia (CAH) is a condition that affects how the body makes hormones. Hormones help the body work and grow. A person with CAH doesn't make enough of a hormone called cortisol (also called the "stress hormone"). There are different types of CAH that can range from very mild to very severe. More testing can help determine which, if any, type of CAH your baby has.



What does this mean?

Although CAH conditions cannot be cured, it can be treated. The most common treatment for CAH are medications that replace the missing hormones that aren't being made correctly. This will help to prevent health problems. If treated early, children with CAH can have healthy growth and development.

What happens next?

Your baby's doctor may ask for your baby to have more testing. You will want to have these follow up tests done as soon as possible. In some cases, you may be asked to visit a specialist called an endocrinologist. The endocrinologist will talk to you about the best plan for your baby.

What are the signs and symptoms of CAH?

If untreated, children with CAH may show signs of early puberty, rapid growth, or no symptoms at all. Some babies might have trouble feeding or gaining weight, or become very sleepy. Girls with classical CAH may have male-looking genitalia. If you become concerned about your baby's growth, feeding or activity, please talk to your pediatrician.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org as an accurate and informative resource.

This fact sheet was written for information purposes only. It should not replace medical advice, diagnosis or treatment.