

Arkansas Department of Health
Newborn Screening Result: Elevated 17-OHP and Positive Steroid Profile
Information Sheet for Parents
Infant may have Congenital Adrenal Hyperplasia

You have just heard that your infant may have Congenital Adrenal Hyperplasia (CAH). Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will have a diagnosis of CAH. Further testing is required to confirm or rule out the diagnosis. The below information is meant to keep you informed while further testing is done to evaluate the newborn screening result.

What is CAH?

CAH is a treatable disorder that occurs when the adrenal glands do not function correctly. CAH is caused by a missing enzyme, causing the adrenal glands to produce too much of some hormones and not enough of others, especially cortisol. The salt-retaining hormone (aldosterone) is another one of those that is under-produced. If left untreated, babies with this condition run the risk of a serious sudden “salt-wasting” episode that can be fatal. A baby with CAH may appear normal at birth, but may have growth and developmental problems.

Why is newborn screening done for CAH?

Newborn screening for CAH provides earlier detection so that treatment can begin earlier. Early detection of CAH is important to preventing many of the complications that may arise should the disorder go untreated during your child’s infancy. In the United States, every one in 15,000 babies is born with CAH.

Does a positive newborn screening result mean that a baby has CAH?

Not always. Some babies who are screened “positive” are found not to have the disorder after further testing. This is because the screening test is not a diagnostic test. A screening test is designed to identify all infants with the disorder, but sometimes picks up infants who don’t have the disorder. Therefore, further testing needs to be done in order to determine if the baby has the disorder, or to rule

it out as a “false positive.” A false positive is more likely if the first heel stick sample was drawn within 24 hours of the baby’s birth or if the baby was premature.

What are the signs and symptoms of CAH?

Deficiency of hormones such as cortisol and the salt-retaining hormone aldosterone may cause generalized weakness and vomiting in newborns. This condition carries the risk of leading to a sudden “salt-wasting” episode, which can be fatal. **Call your baby’s doctor if any of the following symptoms appear: a lot of vomiting, poor weight gain or if your baby looks ill.**

How is CAH diagnosed?

After receiving a positive newborn screen, the most important thing parents can do is be sure that their baby goes in for a new specimen to be collected and tested as soon as possible. A pediatric endocrinologist will help confirm a diagnosis and start treatment.

Where can I get additional information?

http://www.arkansas.gov/newborn_screening/index.html

