

PARENT INFORMATION about INITIAL INCONCLUSIVE Newborn Screening Results for CONGENITAL ADRENAL HYPERPLASIA (CAH)

When your baby was born a small blood sample was taken from your baby's heel to test for certain diseases. These diseases usually cannot be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were "inconclusive" for a disorder called congenital adrenal hyperplasia (a-DREE-nuhl HIGH-per-PLAY-zia). It is important to remember this is only a screening test. A new blood sample will be collected from your baby and tested to find out if your baby may have CAH or not.

What does an "inconclusive" screen mean?

A small percent of all babies screened will be identified as "inconclusive" on screening. Most are later found not to have the disorder. A repeat screening test should be done to determine if your child has this disorder. Keep in mind that the new test could show that your baby does not have CAH.

What is the next step?

Be sure to follow your baby's doctor's instructions for getting a new sample for further testing. CAH is a rare disease involving the endocrine system, so your baby's doctor might consult with or refer you to a pediatric endocrinologist.

What exactly is CAH?

CAH is a rare disease caused by a missing enzyme. When this enzyme is missing, the adrenal glands produce too much of some hormones and not enough of others. The salt-saving hormone is one that is under-produced. Therefore babies with this condition are at risk of a serious sudden "salt-wasting" episode that can be fatal. A baby with CAH may appear normal at birth.

Be sure to call your baby's doctor if any of these symptoms appear: vomiting, poor weight gain or if your baby looks ill.

There can also be other forms of CAH with less serious but still significant symptoms. Children will usually develop normally if treatment begins in the first weeks of life. Finding out early if your baby has CAH, means you and your baby's doctor can take steps to prevent these effects.

What should I do?

The most important thing that you can do is to be sure your baby has a new blood sample collected and tested without delay.

If your baby does have CAH, there are very effective treatments available. Your baby's doctors will be able to help you manage this through checking the hormone blood levels and adjusting the medication to help keep them normal. Treatment should NOT be started before collecting the new blood sample. NO treatment should be started unless a physician recommends it.

Remember, this could be a false alarm. If it turns out your baby does have CAH, the treatment is very effective.

You have probably already done this, but now is also a good time to be sure your baby is covered by your health insurance.

For more information, talk with your baby's doctor. You can contact the **Nebraska Newborn Screening Program at (402) 471-0374** for information on specialists available in Nebraska. Good websites for more information are at www.caresfoundation.org or <http://www.dhhs.ne.gov/nspl/>

Department of Health & Human Services

