

# **PARENT INFORMATION about REPEAT INCONCLUSIVE Newborn Screening Results for CONGENITAL ADRENAL HYPERPLASIA (CAH)**

The results of your baby's repeat screening test continue to be "inconclusive" for congenital adrenal hyperplasia (a-DREE-nuhl HIGH-per-PLAY-zia). The Nebraska Newborn Screening Program recommends that your baby be seen by a pediatric endocrinologist for further evaluation.

## **If the results are still inconclusive, why did we repeat the first test?**

Most of the baby's that have inconclusive results on the first test will have normal results on the repeat test. Repeat testing is less expensive and is easier for most families.

A small number will continue to have inconclusive results on the repeat test. When this occurs, these babies are at an increased risk of having one of the milder forms of CAH. Evaluation by a pediatric endocrinologist can help sort out the problem. The new testing could show that your baby does not have CAH.

## **What is the next step?**

Be sure to follow your baby's doctor's instructions. She/he may recommend getting a new blood sample, or refer you directly to a specialist.

## **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. 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 immediately if your baby has 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. Treatment is also available for these other forms of CAH. Children will usually develop normally if treatment begins in the first weeks of life. Finding out early if your baby has CAH, means steps can be taken to prevent problems.

## **What should I do?**

**The most important thing that you can do is to follow the recommendation of your baby's doctor. Be sure to keep all appointments made with the doctor and the specialist.** If your baby does have CAH, there are very effective treatments available. An endocrinologist will be able to help you manage this by checking the hormone levels and adjusting the medication to help keep them normal.

Treatment should NOT be started before collecting any additional blood samples that are needed. NO treatment should be started unless a physician recommends it. Remember, this could be a false alarm. If 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 information are: [www.caresfoundation.org](http://www.caresfoundation.org) or <http://www.dhhs.ne.gov/nsp/>

Department of Health & Human Services

