

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

The results of your baby's repeat screening test for congenital adrenal hyperplasia (a-DREE-nuhl HIGH-per-PLAY-zia) are positive. The first and second test measured the amount of the steroid 17 hydroxyprogesterone (17OHP). Since the second test shows the level of 17OHP is above normal, your baby needs further testing. The Nebraska Newborn Screening Program recommends that your baby be seen by a pediatric endocrinologist for further evaluation.

## **Why did we have to repeat the 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. Usually the repeat test is all that is needed.

A small number of these babies will continue to have high levels of 17OHP on the repeat test. When this occurs, these babies are usually sent to a pediatric endocrinologist for evaluation. A specialist can help sort out the meaning of the test results. 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 your baby directly to a specialist.**

## **What exactly is CAH?**

CAH is a disease caused by a missing enzyme. When this enzyme is missing, the adrenal glands make 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 important symptoms. 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 these effects.

## **What should I do?**

**The most important thing to do is to make sure your baby has a new blood sample collected and tested without delay.** If your baby has CAH, there are very effective treatments available. A pediatric endocrinologist will be able to help you manage this by checking the blood levels and adjusting the medication to help keep the hormone levels normal.

Medications to treat CAH should NOT be started before the new blood sample for the additional tests is collected. NO treatment should be started unless a physician recommends it.

Remember, this could be a false alarm. If your baby has CAH, the treatment is very effective.

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: [www.caresfoundation.org](http://www.caresfoundation.org) and <http://www.dhhs.ne.gov/nsp/>

Department of Health & Human Services

