

PARENT INFORMATION for INITIAL POSITIVE newborn screening results for CONGENITAL ADRENAL HYPERPLASIA (CAH)

When your baby was born a small sample of blood 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 "positive" 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 a new test will be done to find out if your baby has CAH or not.

What does a "positive" screen mean?

A positive test means that your baby needs to have a new blood test to check for CAH. A small percent of all babies screened will be identified as "positive" on screening, but are later found not to have the disorder. This is just a "screening" test and further testing should be done to determine if your baby has CAH or not.

What is the next step?

Be sure to follow your baby's doctor's instructions for immediately getting a new blood 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 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. **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 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 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 take your baby to have the new blood sample collected and tested immediately. If it turns out your baby does have CAH, there are very effective treatments available, and a pediatric endocrinologist will be able to help you manage this through 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, but if it turns out your baby does have 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 at www.caresfoundation.org or www.magicfoundation.org

