

**Arkansas Department of Health
Newborn Screening Result: Elevated Methionine
Infant may have Homocystinuria
Information Sheet for Parents**

You have just heard that your infant may have Homocystinuria (HCU). Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will be confirmed to have the diagnosis. The below information is meant to keep you informed while further testing is done to evaluate the newborn screening result.

What is HCU?

HCU is a treatable disorder affecting the way the body processes protein. Children with HCU cannot use the amino acid methionine. If left untreated, methionine, homocysteine and other related molecules build up in the bloodstream and may cause brain damage and other disabilities.

Why is newborn screening done for HCU?

Newborn screening for HCU offers early detection so that treatment can begin earlier. Early detection of HCU is key to preventing many of the complications that may arise should the disorder go untreated during a child's infancy.

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

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."

How common is HCU?

HCU affects one in every 300,000 babies.

What are the signs and symptoms of HCU in an infant?

A newborn will usually not have any symptoms.

How is HCU 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.

How is HCU treated?

HCU is treated at first by changing the baby to a diet that restricts methionine and has extra cysteine (another amino acid). Large doses of vitamin B6 can also sometimes be beneficial. Individuals must follow the special diet for the rest of their life.

Where can I get additional information?

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

