

Arkansas Department of Health
Newborn Screening Result: Elevated Citrulline
Infant may have Urea Cycle disorder
Information Sheet for Parents

You have just heard that your infant may have a urea cycle disorder. 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.

Overview

Proteins are made up of building blocks called amino acids. Ammonia is a compound that is formed when amino acids are broken down for use in the body. The body normally changes ammonia into a substance called urea. Urea can then be safely removed in the urine. However, when ammonia cannot be changed into urea, high levels build up in the blood, which is harmful. Affected babies usually do not show signs of the disorder in the first few days of life.

There are two types of urea cycle disorders with increased citrulline – citrullinemia I and argininosuccinic aciduria. Infants with either of these two conditions cannot remove ammonia from the body. If left untreated, ammonia builds up in the bloodstream and may cause brain damage and other disabilities.

Why is newborn screening done for these urea cycle disorders?

Newborn screening offers early detection so that treatment can begin earlier. Early detection is key to preventing many of the complications that may arise.

Does a positive newborn screening result mean that a baby has a urea cycle disorder?

Probably, though some babies who are screened will be identified as “positive” on screening, but

later found to not have the disorder. 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.”

What are the signs and symptoms of a urea cycle disorder?

Symptoms may include poor appetite, extreme sleepiness, irritability, and vomiting. If these signs continue without treatment, the high levels of ammonia may cause decreased muscle tone, breathing problems, problems staying warm, seizures, brain swelling, and coma (which may lead to death). Other effects can include mental retardation, enlarged liver, and poor growth.

How is a urea cycle disorder 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. If the baby does have a urea cycle disorder, there are treatments available.

How is a urea cycle disorder treated?

The baby’s primary doctor will work with a metabolic doctor and a dietician to treat the condition. First the baby’s formula is changed to one that is low in protein. Not all protein is removed from the child’s diet, because protein is essential for normal growth and development. Any changes in diet should be made under the guidance of a metabolic specialist. 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