

Arkansas Department of Health
Newborn Screening Result: Elevated Leucine
Infant may have Maple Syrup Urine Disease
Information Sheet for Parents

You have just heard that your infant may have Maple Syrup Urine Disease (MSUD). **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

Maple Syrup Urine Disease is an inherited disorder in which the body is unable to process certain protein building blocks (amino acids) correctly. The condition begins in early infancy, and involves poor feeding, vomiting, lack of energy, seizures, and developmental delay. Affected infants' urine has a distinctive sweet odor, similar to burned caramel. MSUD can be life-threatening if left untreated.

What is MSUD?

MSUD is a treatable disorder affecting the way the body processes protein. Children with MSUD cannot use specific amino acids (protein "building blocks"). If left untreated, these amino acids build up in the bloodstream, become toxic, and cause brain damage.

Why is newborn screening done for MSUD?

Newborn screening for MSUD offers early detection so that treatment can begin earlier. Early detection of MSUD is important for preventing many of the complications that may arise should the disorder go untreated during a child's infancy. Specialists will be able to improve the quality of life for individuals with MSUD.

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

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."

How common is MSUD?

MSUD affects one in every 185,000 babies. The occurrence is about 1 in 358 among the old order Mennonite population.

What are the signs and symptoms of MSUD?

Babies with MSUD show symptoms as soon as they are fed, usually shortly after birth. Some of the first symptoms include: poor appetite, weak suck, weight loss, high pitched cry, and urine that smells like maple syrup or burnt sugar. Babies can then have episodes of illness called metabolic crises. These signs include: extreme sleepiness, sluggishness, irritable mood, and vomiting. If not treated, further symptoms may follow such as: muscle tone alternating between rigid and floppy, swelling of the brain, seizures, high levels of acidic substances in the blood, and coma sometimes leading to death.

How is MSUD 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 MSUD, there are treatments available.

Is there a cure for MSUD?

There is not currently a cure for MSUD. However, treatment programs are allowing most people with MSUD to live fairly normal lives.

How is MSUD treated?

MSUD is treated with a special diet low in these toxic amino acids. To prevent mental retardation, treatment must begin soon after birth. Because protein is essential for normal growth and development, the child must continue on a diet that provides protein and other nutrients but limits the amount of toxic amino acids. People with MSUD need specialized treatment through a clinic with experience in treating this disorder. 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

