

Positive Result:

Blood Spot Screen Result Notification



Elevated Leucine, Isoleucine, and Other Branched-Chain Amino Acids

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of amino acids called leucine and isoleucine. Other branched-chain amino acids were elevated as well.

What does this mean?

High levels of these amino acids can indicate that your child has a condition called maple syrup urine disease (MSUD). A positive result does not mean your baby has MSUD, but more testing is needed to know for sure.

What happens next?

Your baby's doctor or a metabolic specialist will help arrange for more testing. Your baby will also be seen by a metabolic specialist.

What is maple syrup urine disease (MSUD)?

MSUD is part of a group of disorders called amino acid disorders. Children with MSUD are unable to break down protein from the food they eat. This causes dangerous amounts of a specific group of amino acids called branched-chain amino acids to build up in the body. Leucine and isoleucine are branched chain amino acids. Too much of these branched chain amino acids can cause health problems and make a person's urine smell like maple syrup.

What health problems can it cause?

MSUD is a lifelong condition. If left untreated, it can cause:

- Lack of energy
- Poor feeding
- High-pitched cry
- Urine that smells sweet like maple syrup
- Vomiting
- Intellectual disabilities
- Seizures
- Coma, sometimes leading to death

Children with MSUD can benefit from prompt and careful treatment.

What treatment options are available?

Treatment for MSUD consists of a special diet that avoids protein. Certain medications and supplements like Thiamine may be prescribed. Early treatment can be life-saving.

Children with MSUD should see their regular doctor and a doctor who specializes in metabolic disorders.

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org