

What is MSUD?

Maple Syrup Urine Disease (MSUD) is a rare metabolic disorder in which the body cannot break down certain proteins found in foods, including breast milk and formula. Because the baby cannot digest the proteins, they build up in the body and can cause damage. The signs and symptoms begin shortly after birth, after the baby starts drinking breast milk or formula. A baby with MSUD can become very ill very quickly. There are many different forms of MSUD and signs and symptoms range from life-threatening to mild.

What causes MSUD?

It is inherited from both parents and cannot be “caught” like a cold or “given” to another child.

How is MSUD found?

Shortly after birth, several drops of blood are taken from a baby’s heel. The dried blood sample is sent to the State Department of Health and Senior Services’ Inborn Errors of Metabolism Laboratory, where it is tested for MSUD and several other conditions. These tests are all part of the State’s Newborn Screening Program. If there is any abnormality, the baby’s doctor and parents are notified.

What does the lab look for?

The lab tests the baby’s blood for a certain type of protein (an amino acid called leucine).

Does an abnormal screening test mean that my baby has MSUD?

Screening tests always need to be confirmed by additional testing and medical evaluation.

IF YOU ARE ASKED TO HAVE YOUR BABY RE-TESTED, ACT QUICKLY AND FOLLOW YOUR DOCTOR’S ADVICE

What will my baby’s doctor do?

The doctor may refer you and your baby to a metabolic/genetic specialist. Your baby’s doctor and the specialist will confirm the diagnosis by doing one or more of the following:

- Get a complete medical history and do a physical exam;
- Look for symptoms of poor feeding, irritability, and vomiting; and
- Get another blood test and test the baby’s urine.

How is MSUD treated?

Lifetime treatment may consist of a special diet of foods with low amounts of proteins and frequent monitoring by your baby’s doctor and the metabolic/genetic specialist.

For more information, contact:

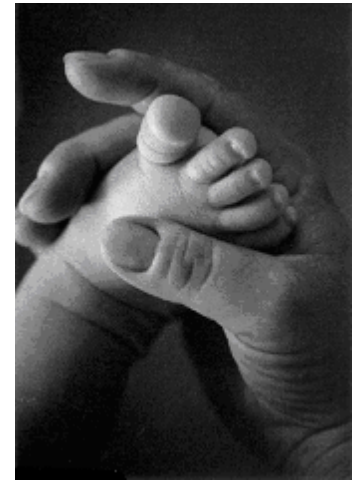
The New Jersey Department of Health
and Senior Services

Newborn Screening and Genetic
Services at (609) 292-1582

or

The Inborn Errors of Metabolism
Laboratory at
(609) 292-3090

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Important Information
For Parents



