

Maple Syrup Urine Disease (MSUD) (Maple syrup urine disease)

What is MSUD?

Maple Syrup Urine Disease (MSUD) is a metabolic disorder. This means the body has a chemical imbalance. MSUD is a condition in which the body cannot break down the essential amino acids leucine, isoleucine, and valine. This causes them to build up in the body. MSUD is inherited. It is present at birth.

What type of problems occurs with MSUD?

A baby who has this condition may have vomiting and poor feeding. If left untreated, this condition can lead to seizures, coma and death. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have MSUD?

This condition occurs in less than 1 in every 100,000 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of MSUD?

The treatment of MSUD is to maintain a special diet with vitamin B (thiamine) added. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care.

Where in Virginia can I take my baby for care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is <http://www.vahealth.org/gns>.



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