

**FACT SHEET**  
Healthcare Provider

**Maple Syrup Urine Disease**

**Description:**

Maple syrup urine disease (MSUD) is an autosomal recessive metabolic disorder. It is caused by a deficiency in branch-chain ketoacid decarboxylation. The result is high body fluid (serum, urine, and spinal fluid) levels of leucine, isoleucine, valine, and their corresponding ketoacids. MSUD has been diagnosed in people worldwide but is most prevalent in the Mennonite population of Lancaster, Pennsylvania.

**Incidence in General Population:**

1:100,000 live births

**Symptoms:**

Affected infants are normal at birth. Within 4 to 5 days, however, symptoms begin to occur.

**Untreated Clinical Features**

- Physical disabilities: Spastic quadriparesis, dystonic posturing, dysarthria, poor physical growth, seizures, central nervous system depression, coma, severe metabolic acidosis, and hypoglycemia.
- Developmental disabilities: irreversible mental retardation.
- Mortality: Lethal usually within two weeks to one month of life.

**Symptomatic Diagnosis**

A symptomatic diagnosis is very possible and should be considered in any infant with severe acidosis in the first 10 days of life. Initial symptoms are poor feeding and marked lethargy along with a characteristic odor of the urine.

**Variants**

There are three variant forms of MSUD: intermediate, intermittent, and thiamine-response forms. All three are associated with deficient decarboxylation of all three branched-chain ketoacids. Physical disabilities range from occasional developmental delay to mental retardation and ataxia.

**Diagnosis:**

Newborn Screening—Tandem mass spectrometry identifies elevations in blood leucine on dried blood spot filter paper. A normal range is 0-277  $\mu\text{mol/l}$ . A second-dried-blood-spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

**Monitoring:**

Individuals diagnosed with MSUD require life-long medical management and dietary therapy coordinated by nutrition and metabolic specialists. Clinical observation is important for healthcare providers caring for patients with MSUD. It is important for primary care provider and the Metabolic Treatment Center to develop an ongoing collaborative relationship in caring for these patients.

**Treatment:**

- Dietary restriction of branched-chain amino acids and supplemental vitamin B1 (thiamine) is essential for a good prognosis.
- During periods of metabolic decompensation, peritoneal dialysis, and/or treatment with intra-venous hyperalimentation without branched-chain amino acids (leucine, isoleucine, and valine) may be necessary.

**Illness and Immunizations:**

- Immunizations should be kept current.
- Consult with the Metabolic Treatment Center within 24 hours of the onset of an illness or at the time of hospitalization.

**Growth and Development:**

Monitor child for normal growth and development.



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