

**MSUD Screening Fact Sheet for Health Care Providers**  
Newborn Screening Program of the Oklahoma State Department of Health

**What is the differential Diagnosis?**

Maple syrup urine disease (MSUD), Hydroxyprolinemia

**What are the characteristics of this amino acid disorders?**

- Autosomal recessive genetic conditions.
- Most infants are born to parents who are both unknowingly asymptomatic carriers and have NO known history of an amino acid disorder in their family.
- The incidence of MSUD is approximately 1:185,000 live births. The incidence among the Mennonite population is 1:760.
- Affected infants appear normal at birth, but usually develop symptoms between **4 and 7 days of life**.
- A maple syrup urine odor may be noted in the urine or cerumen. Newborns present with feeding intolerance, lethargy and vomiting. Untreated, MSUD will quickly progress to cerebral edema, seizures, coma, irreversible mental retardation and possibly death.
- Lifelong treatment includes a restriction of branched chain amino acids and supplementation with medical formula as well as special care during times of illness or stress.

**What is the screening methodology for MSUD?**

1. An amino acid profile by Tandem Mass Spectrometry (MS/MS) is performed on each filter paper.
2. Leucine is the primary analyte.
3. If Leucine is elevated then Valine and the Leucine/Phenylalanine ratio will also be evaluated.

**What is an in-range (normal) screen result for MSUD?**

Leucine < 300 µmol/L is NOT consistent with MSUD.  
See Table 1.

**What is an out-of-range (abnormal) screen for MSUD?**

Leucine ≥ 300 µmol/L requires further testing.

**What screen results will require diagnostic testing?**

All out-of-range leucine screens require **immediate** action.  
The follow-up program will provide detailed guidance on required actions and an *Emergency Management Protocol* will be provided.

<b>Primary Analyte</b>	<b>In-Range (µmol/L)</b>	
Leucine	<	300
<b>Secondary Analytes<sup>1</sup></b>		
Valine	<	280
Leucine/Phenylalanine Ratio	<	4.8

<sup>1</sup> Elevations of the secondary analytes are reported as "not consistent with an amino acid disorder" if the primary analyte is in-range.

**What are the follow-up needs?**

The follow-up program will provide detailed guidance on needed actions. The following metabolic specialists have approved all recommendations:

Metabolic Specialist in Oklahoma

Klaas Wierenga, M.D. (405) 271-8685

Susan Palmer, M.D. (405) 271-8685

**What is my role in screening?**

If you are listed as the infant's planned health care provider on the filter paper requisition, you are required by the *Newborn Screening Program Regulations* to initiate follow-up activities.

Newborn Screening Program (405) 271-6617 or 1-800-766-2223  
Metabolic Nurse Specialist (405) 559-1378 (pager)  
<http://nsp.health.ok.gov>