

## Physician Action Sheet for Abnormal Newborn Screen – Maple Syrup Urine Disease (MSUD)

**Newborn Screen Findings:** Elevated leucine & valine levels on newborn screen, indicating possible maple syrup urine disease (MSUD).

The Indiana University NBS Lab should have already contacted your office & the infant's birthing facility about the abnormal results & recommended confirmatory testing. The IU NBS Lab has also notified the Metabolic Specialist. *Please note that the IU NBS Lab does not contact the patient's family.*

### Required Actions

1. **Contact the Metabolic Specialist (317) 274-3966 (ask for Newborn Screening Nurse or Genetic Counselor) to discuss abnormal results, follow-up recommendations & confirmatory testing.**
2. **Contact the family as soon as possible.**
  - a. Inform the family of the abnormal NBS result & the need for additional confirmatory testing.
  - b. Determine the condition of the infant. *See the infant in your office immediately if there are concerns of poor feeding, lethargy, vomiting, tachypnea, alternating hypertonia/hypotonia or seizures. If symptomatic, see the patient immediately or transport to hospital for further treatment in consultation with the Metabolic Specialist.*
  - c. If patient is not symptomatic, review the list of symptoms that require immediate contact to your office (see above).
3. **Arrange for confirmatory laboratory testing at the infant's birthing facility.**
4. **Plan for follow-up with the Metabolic Specialist if MSUD is confirmed.**

**Clinical Summary:** Autosomal recessive disorder of branched chain amino acid metabolism. Deficient activity of branched-chain ketoacid dehydrogenase (BCKAD) results in elevation of leucine, isoleucine & valine, as well as accumulation of abnormal urine organic acid metabolites. These abnormal metabolites cause metabolic acidosis, poor feeding, vomiting, lethargy, hypotonia/hypertonia, seizures & possible progression to coma. There is a characteristic maple syrup odor to urine & perumen. There are also milder intermittent & thiamine-responsive forms of MSUD.

**Treatment:** Individuals with MSUD require special formula & a diet low in branched chain amino acids & may need special medication or supplements. Treatment is life-long & will require follow-up with Metabolic Specialist. Early diagnosis, preventative care & prompt treatment during metabolic crises will help ensure the best outcome for individuals with MSUD.

**Incidence:** Rare; 1/469,000 in Indiana (1/200,000 nationally). More common in the Mennonite population (1/760). Genetic testing available.

### **Contact Information:**

- Regular business hours: (317) 274 – 3966; ask for the Newborn Screening Nurse or Genetic Counselor.
- Emergency night/weekend contact: (317) 944 – 5000; have the on-call Metabolism physician paged.

### **Resources:**

- Newborn Screening ACT sheets – [www.acmg.net](http://www.acmg.net) (select "Resources," click on "ACT Sheets")
- Information for clinicians - <http://www.ncbi.nlm.nih.gov/sites/GeneTests/review>