

Physician Action Sheet for Abnormal Newborn Screen – Amino Acid Disorders

Newborn Screen Findings: Elevated amino acid levels on newborn screen in a pattern suggestive of an amino acid (AA) disorder. Abnormal metabolites will vary depending on possible disorder.

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 an amino acid disorder is confirmed.**

Clinical Summary: Autosomal recessive disorders of amino acid metabolism. Each AA disorder is associated with deficiency of a different enzyme. *Indiana's newborn screening panel includes homocystinuria, maple syrup urine disease (MSUD), phenylketonuria (PKU), tyrosinemia, citrin deficiency & some urea cycle disorders (citrullinemia & arginosuccinic aciduria).* Symptoms will vary & may present acutely in the newborn period (metabolic acidosis, poor feeding, vomiting, lethargy, hypotonia & seizures with progression to coma & death) or be slowly progressive (resulting in developmental delay, failure to thrive or organ dysfunction). Immediate contact with the Metabolic Specialist & prompt follow-up & confirmatory testing are important for the best outcome.

Treatment: Individuals with AA disorders require special formula & a low-protein diet & may need special medication or supplements. Treatment is life-long & will require follow-up with Metabolic Specialist. Intercurrent illnesses associated with poor intake, vomiting or fever may cause metabolic crises requiring hospitalization. Early diagnosis, preventative care & prompt treatment during metabolic crises will help ensure the best outcome for individuals with AA disorders.

Incidence: Dependent on the specific disorder. 1/7,800 in Indiana (total incidence of all AA disorders on Indiana's newborn screening panel). Genetic testing available for many of the disorders.

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 (select "Resources," click on "ACT Sheets")
- Information for clinicians - <http://www.ncbi.nlm.nih.gov/sites/GeneTests/review>