

Physician Action Sheet for Abnormal Newborn Screen – Phenylketonuria (PKU)

Newborn Screen Findings: Increased phenylalanine & increased phenylalanine:tyrosine ratio on newborn screen.

The Indiana University Newborn Screening (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.** Inform the family of the abnormal NBS result & the need for additional confirmatory testing.
3. **Work with the Metabolism Clinic to set up an appointment for confirmatory testing/follow-up.** Confirmatory testing/follow-up will occur at the Metabolism Clinic at Riley Hospital for Children's Outpatient Center in Indianapolis, IN. Babies will be seen within a few days of the abnormal PKU screen. Confirmatory testing results can be expected one to two days after the child's visit.
4. **Plan for follow-up with the Metabolic Specialist if PKU is confirmed.**

Clinical Summary: Autosomal recessive disorder of amino acid metabolism resulting from deficient activity of phenylalanine hydroxylase. Deficient activity of phenylalanine hydroxylase leads to elevated phenylalanine levels. If untreated, symptoms of PKU appear later in infancy & can include irreversible psychomotor delay, mental retardation, seizures, autistic-like behavior & hyperactivity. A "mousy" or "musty" body odor, pigment dilution & eczema-like rash may also be present with untreated PKU.

Treatment: Treatment will be started promptly after diagnosis under the direction of a Metabolic Specialist. Treatment includes formula & diet low in phenylalanine. Older children may have special medications added to help control phenylalanine levels. Treatment is life-long & will require follow-up with Metabolic Specialist. Early diagnosis, prompt treatment & continued adherence to low-phenylalanine diet will help ensure the best outcome for individuals with PKU.

Incidence: 1/13,000 in Indiana (1/10,000 to 1/25,000 nationally). PKU is more prevalent in Caucasian individuals, although it occurs in all races. 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 (select "Resources," click on "ACT Sheets")
- Information for clinicians - <http://www.ncbi.nlm.nih.gov/sites/GeneTests/review>