

Phenylketonuria (PKU)

Action required

Contact metabolic specialist today.
See infant today.

Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or metabolic clinic
 - § Plasma amino acids
 - 0.5 mL frozen plasma in a sodium heparin green top tube
 - § Urine bipterin
 - § DHPR assay

False Positives

- Common
- Screening result can be impacted by administration of TPN
- Mild variants are seen

Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, lab work, and referral to metabolic clinic. Expect infant to be stable when family is contacted and at clinic visit.

Prompt follow-up is important.

NICU issues

Newborn screens cannot be accurately interpreted after administration of TPN.

Clinical summary

Phenylketonuria (PKU) is an autosomal recessive disorder that results from the defective activity of phenylalanine hydroxylase, the enzyme responsible for breaking down phenylalanine. Newborns are asymptomatic. If an infant is not screened and/or left untreated, symptoms begin to appear later in infancy and can include mental retardation, hyperactivity, autistic-like features, and seizures.

Affected children require life-long dietary restriction of phenylalanine and monitoring by both primary care and specialty providers.

Incidence: ~ 1/15,000; more prevalent in Caucasian individuals.

Clinical expectations

If treated promptly, children with PKU can be expected to be clinically stable and have near normal development.

Developmental delay can occur even with treatment.

Affected children should be monitored for dietary compliance and need for intervention.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening