



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: INCREASED PHENYLALANINE

CONDITION: PHENYLKETONURIA (PKU)

DIFFERENTIAL DIAGNOSIS: Phenylketonuria (Classical PKU); non-PKU mild hyperphenylalaninemia; pterin defects; transient hyperphenylalaninemia.

METABOLIC DESCRIPTION: In PKU the phenylalanine from ingested protein cannot be metabolized to tyrosine because of deficient liver phenylalanine hydroxylase (PAH). This causes elevated phenylalanine. Pterin defects result from deficiency of tetrahydrobiopterin (BH4), the cofactor for PAH and other hydroxylases. This produces not only increased phenylalanine but also neurotransmitter deficiencies.

ACTION TO BE TAKEN IMMEDIATELY:

- ◆ Contact the family immediately to inform them of the newborn screening result.
- ◆ Consult with a pediatric metabolic specialist.
- ◆ Evaluate the newborn and refer as appropriate.
- ◆ Initiate confirmatory/diagnostic tests in consultation with a metabolic specialist.
- ◆ Provide the family with basic information about PKU and dietary management.
- ◆ Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Plasma amino acid analysis which shows increased phenylalanine without increased tyrosine (increased phenylalanine/tyrosine ratio). Urine pterin analysis and red blood cell DHPR assay will identify pterin defects. Consider PAH mutation testing.

CLINICAL EXPECTATIONS: Asymptomatic in the neonate. If untreated, PKU will cause irreversible intellectual disabilities, hyperactivity, autistic-like features, and seizures. Treatment will usually prevent these symptoms. Pterin defects cause early severe neurologic disease (developmental delay/seizures) and require specific therapy.

REPORTING: Report diagnostic results to the family and Kansas NBS program.

SPECIALISTS:

Bryce Heese, MD
Children's Mercy Hospital
Kansas City, MO
Clinic phone: 816-234-3771
Hospital Operator: 816-234-3000

Siddharthan Sivamurthy, MD
KU Wichita Pediatrics
Wichita, KS
Office: 316-962-7386