NEBRASKA
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DEPT. OF HEALTH AND HUMAN SERVICES

Physician ACT SHEET for POSITIVE Newborn Screening Result for PKU (Phenylketonuria)

Disease Category: Amino Acidopathy

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Immediate consultation with a metabolic specialist**
- **Contact family to inform them of the newborn screening result and arrange a visit for an immediate physical exam of the newborn.**
- **Undertake definitive investigations in consultation with metabolic specialist and refer as indicated.**
- **Report findings** to state newborn screening program.

Pediatric metabolic specialists are available through the metabolic centers at Children's Hospital and Nebraska Medical Center. The pediatric metabolic specialist on service/on call today is: _____MD and can be paged at: _____

Meaning of the Screening result: Abnormal or significantly abnormal (elevated) concentrations of phenylalanine or elevated phenylalanine and reduced levels of tyrosine with increased phenylalanine: tyrosine ratio suggests PKU. Elevated phenylalanine can be associated with disorders other than PKU.

Metabolic Description: This autosomal recessive genetic condition is caused by an enzyme defect that impairs the breakdown of an amino acid, phenylalanine into its product, tyrosine because of a deficiency in the enzyme phenylalanine hydroxylase (PAH).

Confirmation of Diagnosis: Specific diagnosis is made by confirmatory tests that show plasma amino acids with increased phenylalanine and decreased tyrosine. It should take no more than 3-4 days to confirm or exclude the diagnosis. **Specimens for "abnormal" or "significantly abnormal" should be collected soon after receiving screening results, no later than 48 hours. Plasma or Serum specimens for quantitative amino acid analysis should be done.**

Clinical Expectations: Asymptomatic in the neonate. If untreated, PKU will produce irreversible mental retardation, hyperactivity, autism, and seizures.