



Newborn Screening ACT Sheet [Increased phenylalanine] Phenylketonuria (PKU)

Differential Diagnosis:

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

Condition 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.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

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

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

Diagnostic Evaluation:

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 Considerations:

Asymptomatic in the neonate. If untreated PKU will cause irreversible mental retardation, 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.

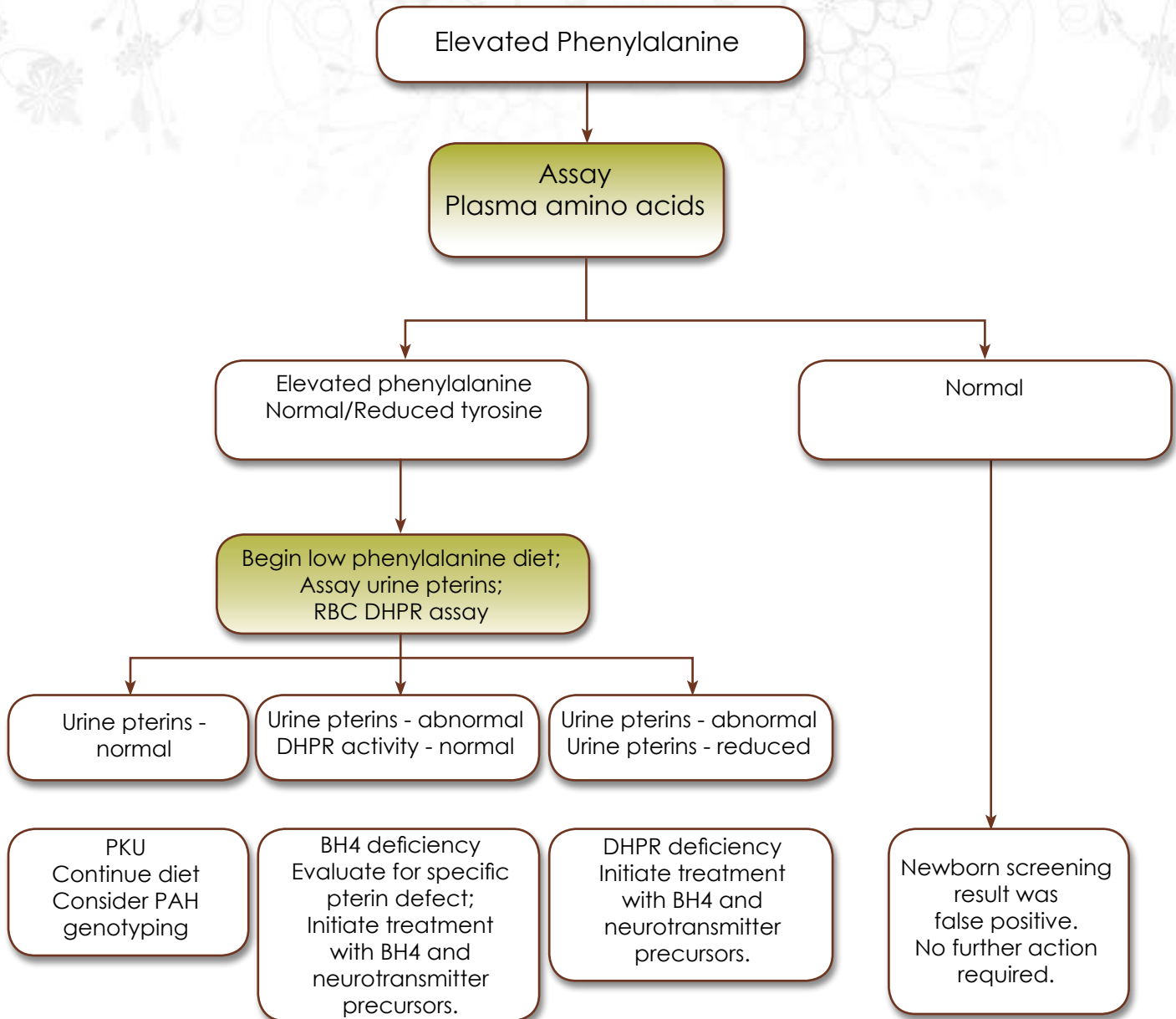
Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen



NEWBORN SCREENING

Screening Today for a Safer Tomorrow

Phenylalanine Elevated



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations / Key:

RBC = Red blood cell
PKU = Phenylketonuria
PAH = Phenylalanine hydroxylase
BH4 = Tetrahydrobiopterin
DHPR = Dihydropteridine reductase

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