Elevated Phenylalanine

Plasma amino acids

Normal

Elevated phenylalanine
Normal/Reduced tyrosine

Begin low phenylalanine diet;
Assay urine pterins;
RBC DHPR assay

Urine pterins - normal
PKU
Continue diet
Consider PAH genotyping

Urine pterins - abnormal
DHPR activity - normal
BH4 deficiency
Evaluate for specific pterin defect;
Initiate treatment with BH4 and neurotransmitter precursors.

Urine pterins - abnormal
DHPR activity - reduced
DHPR deficiency
Initiate treatment with BH4 and neurotransmitter precursors.

Newborn screening result was false positive.
No further action required.

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

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.