Suggested Follow-up for Phenylketonuria
Elevated Phenylalanine (PHE)

Possible Causes: Elevated phenylalanine (PHE) is the primary marker for classical phenylketonuria (PKU). This disorder is caused by decreased activity of phenylalanine hydroxylase. Screening can also identify benign hyperphenylalaninemia and defects in biopterin cofactor biosynthesis or regeneration.

Next Steps if Abnormal: Repeat amino acid profile on filter paper and send to the DHEC laboratory. No formula/feeding changes until result of repeat testing known. If PHE still elevated in repeat specimen, refer to metabolic specialist. Further diagnostic evaluation may be necessary to rule out BH₄ defects. Initiate PHE restricted diet in coordination with metabolic dietitian.

Neonatal Presentation: None.

Emergency Treatment: None.


Advice for Family: Provide basic information about PKU. The handout, When Baby Needs a Second Test for PKU, may be used for this purpose.

Internet Resources:

http://www.pkunews.org/

http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html


http://ghr.nlm.nih.gov/condition=tetrahydrobiopterindeficiency

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