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 BH_4 defects. Initiate PHE restricted diet in coordination with metabolic dietitian.

Neonatal Presentation: None.

Emergency Treatment: None.

Standard Treatment: PHE restricted diet for life. BH_4 defects require additional diagnostic evaluation and treatment.

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=phenylketonuria

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

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