Suggested Follow-up for Phenylketonuria
Elevated Phenylalanine (PHE)

Possible Causes:

Elevated phenylalanine (PHE) is the primary marker for **Phenylketonuria (PKU).** This disorder is caused by decreased activity of phenylalanine hydroxylase.

Screening can also identify **benign hyperphenylalaninemia** and defects in **tetrahydro-biopterin (BH_4) cofactor biosynthesis or regeneration**.

Next Steps if Abnormal:

Repeat amino acid profile on filter paper and send to the DHEC Public Laboratory. No formula/feeding changes are needed until results of repeat testing are known. If PHE is still elevated in the repeat specimen, refer to a metabolic specialist.

Further diagnostic evaluation may be necessary to rule out BH_4 defects. Initiate PHE restricted diet in coordination with a metabolic dietitian and/or specialist.

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://ghr.nlm.nih.gov/condition=phenylketonuria

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

http://www.newbornscreening.info/Parents/aminoaciddisorders/PKU.html#1

https://pkunews.org/

https://www.acmg.net/PDFLibrary/Phenylalanine.pdf