Suggested Follow-up for Homocystinuria
Elevated Methionine (MET)

Possible Causes:

Elevated methionine (MET) is the primary marker for **Homocystinuria.** This disorder is primarily caused by a deficiency in the enzyme cystathionine synthetase. Untreated infants are at risk for developmental delay, intellectual disability, dislocated ocular lens, thromboembolism, and myopia.

Screening for homocystinuria may also identify infants with **Hypermethioninemia**. Primary hypermethioninemia that is not caused by other disorders, liver disease or excess methionine intake appears to be extremely rare.

Next Steps if Abnormal:

See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include plasma amino acids, total plasma homocysteine and urine amino acids.

In addition, **repeat amino acid profile** on filter paper and send to the DHEC Public Health Laboratory.

Neonatal Presentation: Usually none.

Emergency Treatment: Usually none.

Standard Treatment:

MET restricted diet for life. Some affected persons are responsive to Vitamin B6 and may not need MET restricted diet. Betaine is often used.

Advice for Family:

Provide basic information about homocystinuria. The handout, *When Baby Needs a Second Test for Homocystinuria*, may be used for this purpose.

Internet Resources:

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

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

http://www.newbornscreening.info/Parents/aminoaciddisorders/CBS.html

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