Suggested Follow-up for Elevated Citrulline (CIT)

Possible Causes:

Elevated citrulline is the primary marker for **Citrullinemia I, Citrullinemia II and Argininosuccinic aciduria (ASA).** They are deficiencies of specific enzymes in the urea cycle.

Next Steps if Abnormal:

Potential medical emergency. 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, urine amino acids and urine orotic acid.

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

Neonatal Presentation:

May show neurological deterioration in first week of life. Poor feeding, vomiting, grunting respiration, tachypnea, and lethargy progress to seizures, coma and death unless quickly treated. Infants are at risk for hyperammonemia crises.

Emergency Treatment:

Treatment of hyperammonemia includes provision of adequate nonprotein calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed. May use IV Na benzoate or Na phenylacetate and IV arginine (ARG). Dialysis may be necessary to lower ammonia level.

Standard Treatment:

High calorie, protein restricted, ARG supplemented diet. Na phenylbutyrate or Na benzoate may be used to help decrease accumulated toxic precursors.

Advice for Family:

Provide basic information about urea cycle disorders. The handout, *When Baby Needs a Second Test for a Urea Cycle Disorder (Elevated CIT)*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

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

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

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

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