Suggested Follow-up for Tyrosinemia Type I, Elevated Succinyl acetone (SUAC)

Possible Causes:

Elevated succinyl acetone (SUAC) is indicative of Tyrosinemia Type I (TYR I)**.** Tyrosine (TYR) may or may not be elevated. This disorder is caused by a deficiency in the enzyme fumaryl-acetoacetate hydrolase (FAH). Untreated infants are at risk for liver failure, jaundice, delayed growth, and eventual hepatocellular carcinoma.

Next Steps if Abnormal:

See infant as soon as possible to ascertain health status. Consult a pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include plasma amino acids, urine succinyl acetone, urine 4-OH phenylpyruvate and urine 4-OH phenyl lactate.

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

Neonatal Presentation: Usually none.

Emergency Treatment: Usually unnecessary.

Standard Treatment:

TYR and PHE restricted diet for life. NTBC (Nitisinone) is also used to inhibit the degradation of tyrosine and the formation of toxic metabolites. Liver transplantation may be indicated.

Advice for Family:

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

NOTE:

Transient Tyrosinemia of the Newborn is the most common amino acid disorder found in infants, especially those who are premature and/or sick. However, prompt repeat screening is needed as a precaution.

Internet Resource:

https://www.newbornscreening.info/Parents/aminoaciddisorders/Tyrosinemia.html

https://ghr.nlm.nih.gov/condition/tyrosinemia

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