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

Possible Causes: Elevated succinylacetone (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 fumarylacetoacetase. Untreated infants are at risk for liver failure, jaundice, growth retardation and eventual hepatocellular carcinoma.

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, urine succinylacetone, urine 4-OH phenylpyruvate and urine 4-OH phenyllactate. In addition, collect specimen on filter paper for repeat amino acid profile and SUAC and send to the DHEC laboratory.

Neonatal Presentation: Usually none.

Emergency Treatment: Usually none necessary.

Standard Treatment: TYR and PHE restricted diet for life. NTBC (Nitisinone) also used to inhibit the degradation of tyrosine and the formation of toxic metabolites. Liver transplantation if 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 Resources:

http://oregon.gov/DHS/ph/nbs/expand.shtml

http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html

http://www.genetests.org/query?dz=tyrosinemia

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