Suggested Follow-up for Tyrosinemia Type II or III, Elevated Tyrosine (TYR)

Possible Causes: Elevated tyrosine (TYR) **without** elevation of succinylacetone (SUAC) can indicate Tyrosinemia Type II or III (TYR II or III)**.** TYR II is caused by a deficiency in the enzyme tyrosine aminotransferase. TYR III is caused by a deficiency in the enzyme 4-OH phenylpyruvate dioxygenase. Untreated infants with TYR II are at risk for eye and skin lesions with neurological problems including developmental delay. The clinical features of TYR III are not well described; however, mental retardation and behavioral problems have been found in affected persons.

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 and urine phenylolic acid metabolites. In addition, collect specimen on filter paper for repeat amino acid profile and send to the DHEC laboratory.

Neonatal Presentation: Usually none.

Emergency Treatment: Usually none necessary.

Standard Treatment: TYR and PHE restricted diet for life.

Advice for Family: Provide basic information about TYR II or III. The handout, *When Baby Needs a Second Test for TYR II or III*, 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.acmg.net/resources/policies/ACT/condition-analyte-links.htm