Suggested Follow-up for Maple Syrup Urine Disease
Elevated Valine (VAL) and/or Elevated Leucine (LEU) + Isoleucine (ILE)

Possible Causes: Elevated valine (VAL) and/or leucine (LEU) + isoleucine (ILE) are the primary markers for **maple syrup urine disease (MSUD).** (LEU and ILE cannot be measured separately in the tandem mass spectrometer.) This disorder is caused by deficiencies in the branched chain keto-acid dehydrogenase complex and leads to accumulation of leucine (LEU), isoleucine (ILE), valine (VAL) and alloisoleucine.

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 and urine organic acids. Presence of alloisoleucine in plasma is key finding for MSUD. In addition, repeat amino acid profile on filter paper and send to the DHEC laboratory.

Neonatal Presentation: May show neurological deterioration in first week of life. Lethargy and poor feeding followed by abnormal muscle tone, involuntary movements, seizures and coma.

Emergency Treatment: Treatment includes provision of sufficient nonprotein calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed.

Standard Treatment: LEU restricted/ILE, VAL controlled diet for life. Some persons with a less severe form of MSUD are thiamin responsive.

Advice for Family: Provide basic information about MSUD. The handout, *When Baby Needs a Second Test for MSUD*, 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://oregon.gov/DHS/ph/nbs/expand.shtml

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

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

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