ACUTE MANAGEMENT OF MAPLE SYRUP URINE DISEASE

- Such patients easily and frequently decompensate with minor infections, poor oral intake, vomiting or constipation. The patient should be admitted immediately to ER and managed as triage level II.
- Immediate actions which should be accomplished within 1 hour of arrival:
- Basic life support
- Stop all source of protein central and parenteral nutrition.
- Check GlucoChecks.
- Insert an IV line and take bloods for the following: blood gas, Chem 1, Ammonia, and urine analysis to check ketones. In addition to plasma amino acids.
- Call Biochemical Genetics lab (Metabolic) lab for urgent processing of plasma aminoacids at **40195, 40196, 40197**
- Start immediately one and half to double maintenance I.V.F as 10% Dextrose + KCL 20 Meq/l. Re-adjust according to lab results (Keep GlucoChecks 5-8mmol/L). Consider start insulin if hyperglycemia develop at dose of 0.01-0.05 unit/kg/hour and titrAate up until blood glucose controlled.
- Start IV intralipid 20% at 2-3 g/kg/day to provide additional calories.
- Call the pharmacy hotline# to expedite the delivery of medications .
- Give prophree or polycose (non-protein calorie source) P.O/NGT as tolerated.
- Identify and treat the infection or other cause of the metabolic stress
- When there is nausea or vomiting give Granisetron 10 to 40 microgram / kg, infused over 3 to 5 minutes.
- Call metabolic dietician on-call to help with dietary management.
- Begin enteral therapy if tolerated with a metabolic formula (Ketonex®-), 2.5-3.5 g/kg/day.
- Begin 1% solutions of Isoleucine and Valine when levels approach upper treatment range (see table). Start supplements at 20-30 mg/kg/day (1.5 to 3.0 mL/kg). Dose range between 20-120 mg/kg/day and adjust based on Plasma amino acids results.
- It is important to realize that isoleucine and valine levels may drop rapidly and that very low levels (isoleucine < 100 umol/L and valine < 200 umol/L) will keep the leucine level from dropping by limiting protein synthesis. Low levels will also allow more leucine to enter brain by providing less transport competition and thus will produce or enhance brain edema and neurological complications.
- Call pharmacy to expedite the intralipid 20% and medications.
- Monitor plasma amino acids and urine for ketones and ammonia daily until patient improved clinically and leucine came back to upper treatment range.
- Consider dialysis if: persistently high plasma leucine despite above measures or leucine encephalopathy or If hyperammonemic.
- Call Biochemical Geneticist (metabolic) on call Tel: 018011111

For more information please read the attached guidelines for this disorder.