Perioperative Management of a Patient with Maple Syrup Urine Disease Undergoing Open Heart Surgery

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Introduction
Maple syrup urine disease (MSUD) is an autosomal recessive metabolic disorder caused by deficiency of the mitochondrial enzyme branched-chain ketoacid dehydrogenase involved in the degradation of branched-chain amino acids (BCAAs; leucine, isoleucine, and valine) and their ketoacid derivatives (BCKAs). Restriction of dietary protein and a specialized diet helps normalize BCAA and BCKA concentrations in blood, limiting long-term effects on brain development. Catabolic states associated with periods of stress such as surgery or infection can lead to acute metabolic encephalopathy and life-threatening brain edema. Literature review identified a limited number of case reports of MSUD patients undergoing surgery, and only one case report detailing management of MSUD after open heart surgery. Careful management of MSUD patients in the perioperative period can decrease the potential for metabolic decompensation.

Case
31 y/o F with MSUD presenting for closure of an atrial septal defect (ASD). The patient was found to have secundum-type ASD during liver transplantation evaluation. The ASD was not amenable to device closure due to multiple fenestrations. The patient was diagnosed with MSUD as a neonate when she presented with neurologic manifestations including opisthotonus. She had moderate intellectual disability and had only been hospitalized a few times due to metabolic decompensation with the last at <10y old. She maintains normal levels of leucine, isoleucine and valine with a restricted diet that includes a metabolic formula and limited protein from table food. A sick day modified formula is utilized when she is ill.

Patient Management
The patient was admitted the day prior to surgical intervention to establish baseline BCAAs and metabolic management by the Medical Genetics Service while NPO. IV fluids with a glucose infusion rate of 8 mg/kg/min was maintained while NPO overnight. For surgery, the CPB circuit was primed with albumin and 2 Units PRBCs to decrease the stress associated with dilutional anemia. Maintenance IV fluids with dextrose were continued throughout surgery, and insulin was titrated to maintain her blood glucose between 150-180 mg/dl. The patient was extubated at the conclusion of the surgery and taken to CICU for close monitoring. The evening following surgery, the patient's regular oral MSUD formula was restarted including valine and isoleucine supplements. On POD #2, there was evidence of increased catabolism with mild increased leucine levels for which the patient was placed on her "sick day" formula, and intralipids were added for additional calories. Leucine levels returned to baseline thereafter. The hospital course was complicated by A-Fib, pleural effusion and a small to moderate pericardial effusion. The patient was discharged on POD#11 and on her follow up visit, a month latter, showed improvement with almost resolution of the effusion. She will follow up with cardiology in 6 months. The patient is currently awaiting liver transplant.

Conclusion
- Acute metabolic decompensation in a patient with MSUD is typically precipitated by a stressor such as infection, injury, surgery, hormonal changes, or significant dietary changes.
- This case illustrates successful perioperative management of a MSUD patient who underwent cardio-pulmonary bypass for ASD closure with close coordination between a metabolic physician, Anesthesiology, Intensive Care, and Surgery.

References
1. Fukutomi M et al. Heart Vessels 1993 8:48-51