Acute Decompensation of Late-Onset Glutaric Acidemia Type II in the Setting of Multifactorial Cirrhosis

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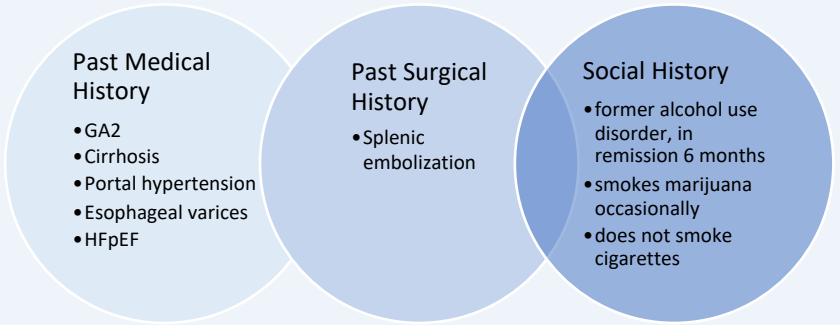
Introduction:

Glutaric acidemia type II (GA2), also known as multiple acyl-CoA dehydrogenase deficiency (MADD), is a disorder that interferes with the body's ability to break down proteins and fats to produce energy. It is due to the disruption of fatty acid oxidation, amino acid and choline metabolism, as GA2 is inherited in an autosomal recessive manner by changes in the ETFA, ETFB, or ETFDH genes involved in electron transfer. The classic and most severe form of GA2 is neonatal onset which is most often identified on newborn screening and can involve metabolic decompensation, organ malformation, and an odor resembling "sweaty feet". Our patient has symptoms of late onset disease seen in a small subset of GA2, including fatigue, vomiting, episodic hyperammonemia, fatty infiltration of the liver, and an absence of congenital malformations. In acute decompensations, triggered by catabolism and/or intercurrent illness, individuals with GA2 may develop metabolic acidosis, rhabdomyolysis, elevation of transaminases, and hyperammonemia.

Case Description:

This case describes a 36-year-old gentleman with late onset GA2, multifactorial cirrhosis from non-alcoholic steatohepatitis in the setting of an inherited metabolic disorder, class III obesity, alcoholic associated liver disease, portal hypertension, splenic embolization, and CHF, that presented to the ED for confusion following progressive nausea and vomiting over five days and found to have elevated an ammonia level at 165. His vital signs were tachycardia of 133 bpm, hypertension of 221/133, afebrile with no respiratory distress. Physical exam was notable for a drowsy obese male oriented to self and place, diaphoretic and tachycardic, no JVD, no crackles, non-tender abdomen with normoactive bowel sounds, and asterixis. Laboratory studies were significant for platelets 131k, K 2.4, CK 2138, total bilirubin 3.7, AST 355 ALT 130 ALP 143, and no detectable ethanol, acetaminophen or salicylate levels. Multiple factors can influence his ammonia level, including underlying cirrhosis and GA2. His metabolic crisis was suspected due to a catabolic state from intake of high-protein diet, and was managed with 10% Dextrose IV fluids, levocarnitine, riboflavin, lactulose, rifaximin, diet low in protein and fat with improvement in encephalopathy. Hypokalemia from decreased absorption and diarrhea from lactulose use resolved with potassium supplements and the addition of spironolactone.



INITIAL LABORATORY STUDIES

Platelets 131K	Creatinine kinase 2138 IU/L	AST 335 IU/L	ALT 130 IU/L	ALP 144 IU/L
Potassium 2.4 mmol/L	Ammonia 165 mcmol/L	pro-BNP 94 pg/mL	Glucose 108 mg/dL HCO3 27 mmol/L	Anion Gap 10 Lactic Acid 1.9 mmol/L

Discussion:

Metabolic disorders are rare but important to consider in patients that present with a metabolic crisis. They can decompensate after periods of decreased oral intake, dehydration, increased exercise, alcohol ingestion or during illness and develop hyperammonemia, transaminitis, metabolic acidosis. These individuals have a lifelong risk of intermittent episodes of metabolic crisis and prompt management is imperative to recovery.