G134 Medium-Chain Acyl-CoA Dehydrogenase Deficiency: A Differential Diagnosis in a Patient With Mental Status Changes Suspected of Drug Toxicity

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The goal of this presentation is to show how rare inborn errors of metabolism such as Medium-Chain Acyl-CoA deficiency can be present with symptoms commonly associated with drug toxicity.

This presentation will impact the forensic science community by showing how patients with nausea, vomiting, and acute mental status change in the setting of fasting should be tested for inborn errors of metabolism such as Medium-Chain Acyl-CoA deficiency if toxicology is negative. When recognized, family counseling can be initiated and additional lives potentially saved.

This case involves a 30-year-old white male coal miner who experienced nausea and vomiting with a 30-pound weight loss over the last three months of his life. He was admitted to the local hospital for a further evaluation of intractable vomiting and complaints of abdominal pain and scant hematemesis. Upon admission, the patient appeared to be in no acute distress and was alert and oriented to person, place, and time. Admission laboratory investigation demonstrated blood glucose of 71mg/dL (70 – 99), BUN 31mg/dL (7 – 18), Creatinine 2.0mg/dL (0.6 – 1.3), and an anion gap of 23.7mEq/L (10 – 20). Several hours into the admission, the patient became confused, anxious, and appeared to be hallucinating with bizarre manifestations, such as licking the air followed by self-injurious behaviors such as banging his head on the wall and trying to jump out a window. He was subsequently placed in soft restraints for his own and others' safety. Fifteen minutes later the patient became unresponsive and developed a rapid respiratory rate (40/min). In another 15 min, respirations and cardiac electrical activity suddenly ceased, and resuscitation attempts were unsuccessful. No ventricular fibrillation was observed during his hospitalization. Drug toxicity, specifically "bath salts," were suspected due to the patient's mental status changes, a history of remote drug abuse in the patient's past, and the prevalence of psychosis-inducing drugs in the community. Drug testing for synthetic stimulants such as mephedrone, MDPV, and methylone were all negative. The past medical history of this patient was unremarkable for previous mental status change. He furthermore had a sibling who had experienced a mild mental status change during a fast for colonoscopy, which was relieved with electrolyte drink and broth.

On autopsy, the patient was a well-developed male with features suggestive of recent weight loss (loose soft abdominal skin and striae). Findings included gross and microscopic microvesicular steatosis, mild cerebral edema, and subnuclear vacuoles of the renal cortical tubules. Autopsy urine and blood were negative for ketones. Postmortem screening of the blood, urine and vitreous were positive only for ethanol and promethazine prescribed in the hospital. Due to the odd presentation of fatty liver, negative drug screen, vomiting, and altered mental status, the forensic pathologist elected to request postmortem metabolic screening of the blood and bile, which was positive. The patient was found to have died due to acute metabolic decompensation caused by Medium-chain acyl-CoA dehydrogenase deficiency, an inherited fatty acid oxidation disorder. Discussed will be the necessity of awareness of such entities as adult manifestation of inborn errors of metabolism in spite of their relative rarity.

MCAD Deficiency, Mental Status Change, Metabolic Disease

 ** Presenting Author*