Anesthesia for Carnitine-Acyl Carnitine Translocase Deficiency

Faizaan Syed, Henry Turner, Faris AlGhamdi, Dmitry Tumin, Joseph Tobias, Tariq Wani
Department of Anesthesiology & Pain Medicine, Nationwide Children's Hospital, Columbus, Ohio

NATIONWIDE CHILDREN'S
When your child needs a hospital, everything matters.

Introduction

- Carnitine and carnitine-acylcarnitine translocase (CACT) play an essential role in the transport of fatty acids into the mitochondria.
- Mutation in the SLC25A20 gene is cause of CACT deficiency.
- Deficiency leads to reduced transport of long chain fatty acids into the mitochondria, thereby limiting use of fatty acids for energy production especially during prolonged fasting, febrile illnesses, increased muscular activity, and other periods of systemic stress.

Clinical symptoms: Neurological and cardiovascular abnormalities, skeletal muscle damage, liver dysfunction, emerging in infancy with high mortality rate.

Metabolic consequences: Hypoketotic hypoglycemia during fasting, hyperammonemia, elevated creatine kinase and transaminases, dicarboxylic aciduria, very low free carnitine and an abnormal acylcarnitine profile.

Clinical presentation: Lactic acidosis, hypoglycemia, hyperammonemia, hepatomegaly with elevated liver enzymes, reduced plasma carnitine levels and skeletal muscle weakness.

We present our anesthetic management of a 10-year-old, 28.8 kg girl with CACT deficiency who presented for left pelvic osteotomy, bilateral varus derotational osteotomies, hip arthrogram, and adductor releases.

Summary

Medical history: CACT deficiency, quadriplegic cerebral palsy, intractable epilepsy, cortical blindness, developmental delay, duplex kidney, glutaric aciduria type II, and gastroesophageal reflux disease.

Surgical history: Corpus callotomy resection, multiple endoscopies, Nissen fundoplication, tonsillectomy and adenoidectomy.

Preoperative examination: Multiple contractures and increased muscle tone.

Preoperative laboratory: Normal electrolytes, hepatic function, coagulation profile, and blood glucose with normal cardiac function on echocardiography.

Admission: A day before surgery, the patient was admitted for PICC line placement for a continuous glucose infusion when she was *nil per os*.

Anesthesia: Planned with concerns for difficult vascular access, blood product use, and invasive monitoring. Anesthetic induction with propofol (3 mg/kg) and maintainance with isoflurane in air/oxygen. Epidural anesthesia with fentanyl and no local anesthetic agent given concerns of sympathectomy and hypotension. Intravenous fluids included 5% albumin and normal saline for replacement fluids with maintenance fluids of D5 Lactated Ringer's.

Intraoperative events: Two episodes of self-resolving bradycardia with a heart rate below 50 beats/min on two occasions with no blood pressure changes. The surgical procedure lasted 8 hours with an estimated blood loss of 150 mL.

Recovery: Uneventful postoperative course. The patient's trachea was extubated following the procedure. She was observed for 24 hours in the ICU and discharged home on postoperative day 6.

Discussion

- CACT deficiency patients warrant preoperative evaluation of neurological status, cardiovascular function, hepatic function, coagulation profile, and blood levels of ammonia and glucose.
- Fasting guidelines need to be modified, and preoperatively, a continuous infusion of dextrose ishould be used to maintain normoglycemia.
- During prolonged surgery, intravenous fluids containing dextrose should be used, with regular intraoperative blood glucose checks. In our patient, a spike in blood glucose after dextrose administration led to its discontinuation with normoglycemia maintained during the procedure.

References

1. Niezgoda J, Morgan PG. Paediatr Anaesth 2013;23:785-93.
2. Pande SV, Parvin R. J Biol Chem 1980;225:2994-3001.
3. Rowe RW, Helander E. Anesth Analg 1990;71:295-7.