[NM-190] Case Report: Anesthesia management in a 4 year old with Merosin-deficient congenital muscular dystrophy undergoing a nephrectomy

Lau J, Matar M, Vustar M
Children's Hospital, Los Angeles , Los Angeles , CA, USA

We present a 4-year-old girl, 29kg, with Merosin-deficient congenital muscular dystrophy (MDCMD) who presented with a ruptured anaplastic Wilm’s tumor. She was born at 28 weeks gestation by c-section due to maternal pre-ecclampsia and spent 2 weeks in the NICU for prematurity and feeding problems without intubation. She was behind in meeting motor development milestones and did not start walking until age 3. After evaluation for delay and frequent falls, she was diagnosed with MDCMD by muscle biopsy. After a fall, the patient complained of abdominal pain and was found to have a ruptured right renal mass and was scheduled for embolization of the right renal artery and radical nephrectomy.

Prior to surgery an echocardiogram was obtained which revealed an EF of 44% and mildly depressed left ventricular wall motion. On physical exam, she had a Mallampati I airway with adequate mouth opening and neck range of motion. She had poor muscle strength and tone in all 4 extremities but otherwise no focal neurologic deficits. Monitored with standard ASA monitors and an arterial line, she was induced and intubated successfully with propofol and rocuronium and maintained on propofol, remifentanil, and dexmedetomidine infusions. Due to the duration of anesthesia and risk for post-op ventilatory weakness, the decision was made to leave the patient intubated. She was monitored overnight in the PICU and extubated without complication the following morning.

Merosin-deficient congenital muscular dystrophy (MDCMD) is a rare heterogeneous subtype of CMD due to mutations in the laminin alpha-2 chain of Merosin, a matrix protein linked with dystrophin. The incidence is estimated to be 0.68-2.5 per 100,000 [1]. Merosin is expressed in cardiac muscle, schwann cells, skin, and skeletal muscle. Clinically this manifests as hypotonia, joint contractures, scoliosis, restrictive pulmonary disease, white matter abnormalities on MRI, and cardiomyopathy. Unfortunately, no known treatment is available. The intubation can be difficult due to jaw contractures and hypotonia. Age at onset is 6-12 months [1,2]. While an increased incidence of MH has not been established in CMD, there is an increased risk for rhabdomyolysis and hyperkalemia and many advocate total intravenous anesthesia [3]. Due to the progressive nature of the disease, patients with MDCMD should have their pulmonary and cardiac function evaluated. Compared with other CMD children, those with MDCMD have a higher incidence of dilated cardiomyopathy [2], making a cardiac work up and echocardiogram prudent. In this case, the patient had no clinical signs or symptoms of cardiac compromise yet still had evidence of LV dysfunction on the study. Understanding the pathophysiology of MDCMD can guide the preoperative work-up and help plan a successful anesthetic plan.

1. Ip et al. Merosin-deficient congenital muscular dystrophy: A case report with MRI, MRS, and DTI findings. J of Radiology 2012, 6(8):1-7.
2. Spyrou, N. et al. Evidence of left ventricular dysfunction in children with Merosin-deficient congenital muscular dystrophy. Am. Heart Journal 1998, 136(3):474-476.
3. Lerman, J. Perioperative management of the paediatric patient with coexisting neuromuscular disease. Br J Anaesth. 2011, 107:79-89.
