Case Study: Spinal anesthesia in an infant with Pompe’s disease (Glycogen storage disease type 2)

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Introduction: Glycogen storage disease type 2 (Pompe’s disease) may present within the first few months of life with generalized hypotonia, muscle weakness and cardiomyopathy. This is an autosomal recessive condition caused by a deficiency of acid alpha glucosidase enzyme resulting in lysosomal glycogen accumulation in the cardiac and skeletal muscles.1 The infantile form has a poor prognosis and death usually occurs by 1 year of age from respiratory or cardiac failure.1,2 Until recently the mainstay of treatment was symptomatic, but enzyme replacement therapy with various forms of recombinant human alpha glucosidase enzyme has improved the outcome of the infantile form.7 During the first year of life, these patients require anesthetic care for diagnostic muscle biopsy and placement of central venous line for enzyme therapy.2 The anesthetic management considerably influences the morbidity and mortality of these infants and regional anesthesia is suggested as an alternative to general anesthesia whenever feasible.3-6 We report the use of spinal anesthesia in an infant with severe non-obstructive cardiomyopathy and respiratory compromise presenting for a diagnostic muscle biopsy which confirmed Pompe’s disease.

Case report: An 8 month-old full term female infant, 6.7 kg, presented for a diagnostic muscle biopsy. She was healthy until 6 weeks prior to the scheduled procedure she developed intermittent fever, weakness, feeding difficulty and 2 weeks later presented to ED with tachypnea, cough and noisy breathing. Physical examination revealed hypotonia, shallow breath, RR 72 bpm, SpO2 99%, audible gallop and enlarged liver. ECG revealed severe ventricular hypertrophy and T-wave inversion in the lateral precordial leads with a strain pattern. ECHO revealed severe concentric LV hypertrophy and dysfunction, EF 38%, mild mitral valve regurgitation and a small pericardial effusion. Chest radiograph demonstrated cardiomegaly causing left lower lobe atelectasis and a moderate left pleural effusion. Medical treatment was deferred pending definitive diagnosis. On the day of surgery, RR was 76 bpm, SpO2 97%, HR 145 bpm, & BP 90/45 mmHg.

Spinal anesthesia was performed after anesthetizing the L3-S1 lumbar skin with EMLA cream. An intravenous access was placed prior to the spinal tap. Standard monitors were applied and maintenance fluid of normal saline was initiated at a rate of 4 cc/kg/h. The lumbar area was prepped and draped aseptically. The spinal tap was performed in sitting position at L5-S1 interspace using a 22-gauge 1.5-inch long spinal needle and clear CSF was obtained. A total dose of 3.5 mg of hyperbaric tetracaine 0.5% with epinephrine 40 mcg was injected uneventfully. The infant was maintained in sitting position for 45 seconds and then was placed in supine position with 20-degree head elevation. Systolic and diastolic BP decreased by 2-3% over the course of the operation and SpO2, HR & RR remained unchanged. The muscle biopsy was completed within 40 minutes. There were no perioperative complications and the infant was discharged home two hours after the end of the surgery.

Discussion: Pompe’s disease is a genetic disease that affects cardiac and skeletal muscles and the untreated infantile type presents unique cardiac and respiratory anesthetic challenges. It is associated with high general anesthesia risks requiring special attention to 1) maintain adequate LV filling volume and coronary perfusion 2) avoid myocardial depression and the potential for dynamic left ventricular outflow obstruction3 3) careful monitoring of myocardial ischemia, arrhythmia with use of 5-lead ECG and 4) weak respiratory muscles that predispose to increased sensitivity to non-depolarizing muscle relaxants, CNS depressants, and may necessitate postoperative ventilation support.6 We elected spinal anesthesia in our patient because it produces satisfactory surgical analgesia, causes minimal hemodynamic changes, avoids tracheal intubation and maintains spontaneous ventilation.
References:
1. Raben, N. Current Molecular Medicine 2002;2:145
2. Ing, R.J. et al., Pediatric Anesthesia 2004;14:514