475840 - MANAGEMENT OF GENERAL ANESTHESIA IN AN INFANT WITH DUPLICATION 3Q SYNDROME AND MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

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Purpose: Duplication 3q (dup(3q)) syndrome is a rare chromosomal aberration with several clinical features of interest to anesthesiologists. These include airway malformations such as micrognathia, cleft palate, and short neck (1,2). Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is the most common of the inherited disorders of fatty acid metabolism, with an estimated incidence of 1:10000 to 1:15000 live births (3). Affected patients are prone to hypoglycemia during periods of fasting, including the perioperative period. We describe the management of a general anesthetic in an infant with concurrent diagnoses of dup(3q) syndrome and MCAD deficiency.

Clinical Features: Consent was obtained from the patient's mother for publication of this case report. A 9 month old female with concurrent unrelated diagnoses of dup(3q) syndrome and MCAD deficiency presented with severe gastroesophageal reflux. She was scheduled for Nissen fundoplication and insertion of feeding gastrostomy tube.

Preoperative investigations showed several airway abnormalities (cleft palate, micrognathia, short neck), but no anomalies in other major organ systems. In consultation with our institution's genetics and metabolic disease team, feeds were held 6 hours before surgery and intravenous glucose supplementation at 10 mg/kg/min was initiated. Despite this, significant hypoglycemia (glucose 2.4 mmol/L) developed after 4 hours of fasting, requiring correction with IV glucose bolus. Because of airway concerns from the preoperative physical examination, spontaneous ventilation was preserved during induction of general anesthesia, using sevoflurane in oxygen by mask and incremental doses of propofol to a total of 3 mg/kg IV. Direct laryngoscopy revealed the presence of a small hypoplastic epiglottis, but endotracheal intubation was otherwise uneventful.

Examination of the patient's back after induction revealed unusual sacral anatomy, precluding the usage of caudal neuraxial analgesia. Glucose levels were closely monitored intraoperatively, while maintaining continuous IV glucose supplementation at 10 mg/kg/min. After the procedure, the patient was successfully extubated and transferred to the intensive care unit for postoperative management. Glucose levels were maintained in the normal range during her ICU stay. Feeds via her gastrostomy tube were started on the third post-operative day. She was transferred to the regular ward on her sixth post-operative day and discharged from hospital on the eighth post-operative day.

Conclusion: This case is the first literature report of anesthetic management in a patient with dup(3q) syndrome, and illustrates airway and neuraxial anomalies as potential anesthetic considerations. Hypoplastic epiglottis was an unexpected finding, and may represent a new congenital defect associated with the dup(3q) syndrome. Finally, hypoglycemia occurring despite the timely initiation of IV glucose supplementation underscores the need for close monitoring during periods of perioperative fasting in MCAD deficiency.

References: 1 Am J Med Genet 2005 134A:434-438 2 Ophthalmic Genetics 2005 26:37-43 3 J Inher Metab Dis 2005 28:141-152