Congenital Catecholamine Deficiency: Anesthetic Management of a Patient with Aromatic L-amino Acid Decarboxylase Deficiency (ALAAD)

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Introduction: Aromatic L-amino acid decarboxylase deficiency (ALAAD) is a rare congenital disorder characterized by absence or near absence of circulating catecholamines. This rare enzyme deficiency leads to autonomic dysregulation, oculogyric crisis and severe developmental delay. The treatment of this rare disorder involves the use of both dopamine agonists and MAOI inhibitors in an attempt to up regulate circulating levels of catecholamines. We are aware of only one report of the anesthetic management of a patient with ALAAD, with less than 40 cases identified worldwide.

Case Report: Our patient, the second of four children, was born 2.1kg at 36 weeks via uncomplicated cesarean section following a diagnosis of oligohydramnios. Mother and daughter were discharged without complications two days following delivery. A few months later she was found to have autonomic instability and poor tone and subsequently her medical history was rapidly expanded to include the diagnosis of ALAAD. Additionally, she suffered from hypotonia, sleep apnea, chronic sinusitis and severe developmental delay. Her autonomic instability was treated with daily midodrine and tranylcypromine. General endotracheal anesthesia was performed at age nine for the placement of a jejunostomy tube secondary to inadequate oral intake. Her anesthetic comprised of 0.5mg/kg oral midazolam sedation followed by 0.3mg/kg intravenous etomidate, vecuronium and isoflurane maintenance. Seven months later this child underwent oral sedation with 0.5mg/kg midazolam and 3mg/kg ketamine for replacement of a cracked jejunostomy tube. Six months later she underwent sedation for a displaced jejunostomy. Sedation during this procedure was accomplished with 0.5mg/kg oral midazolam.

Discussion: An inherited deficiency of aromatic L-amino acid decarboxylase leads to profoundly diminished or absent circulating catecholamines. The anesthetic implications of this enzyme deficiency are multifaceted and involve hemodynamic regulatory dysfunction, heart rate variability and labile blood pressure regulation. In contrast with intrinsic sympathetic nervous system dysfunction, patients with ALAAD deficiency have an intact parasympathetic system leading to unbalanced vagal outflow. This unbalanced vagal outflow may result in ‘dystonic spells’ bradycardia, hypotension and lack of compensatory response to blood loss. The rationale behind sedation for jejunostomy replacement in our patient was to reduce the autonomic instability during episodes of increased stress, agitation and pain. This report illustrates multiple anesthetic...
techniques employed in a child with a very rare enzyme deficiency in an effort to shed insight into the management of this rare disease process.

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