

Anesthesia Management Of A Patient With Williams Syndrome

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Citation

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Abstract

Williams syndrome is a genetic disorder characterized by developmental delay, unusual facial appearance, narrowing of the aorta (large artery that leaves the heart) and particular cognitive and personality profiles. Chronic abdominal pain is a common complaint of children and adults with Williams syndrome; possible causes include hiatal hernia, peptic ulcer disease, cholelithiasis, diverticulitis, ischemic bowel disease, chronic constipation, and somatization of anxiety. A 5-month-old male infant, weighing 4.7 kg with Williams syndrome was scheduled for inguinal hernia repair under general anesthesia. The diagnosis of Williams syndrome was made at 4 months of age. Intraoperative and postoperative course was uneventful. We think that general anaesthesia can be performed successfully with hemodynamic stable

INTRODUCTION

Williams syndrome (WS) is an uncommon genetic syndrome due to a deletion of several genes on chromosome 7 that includes the elastin gene. The syndrome is associated with dysmorphic facies, neurological manifestations, idiopathic hypercalcemia, and cardiac abnormalities, particularly supravalvular aortic stenosis (SVAS) (1). Williams syndrome was first described by Williams, Barratt- Boyes, and Lowe in 1961 (2). The problem is usually caused by a random mutation, so parents may not have any family history of the condition. However, a person with Williams syndrome has a 50% chance of passing the disorder on to each child. It occurs in about 1 in 20,000 births (3). This is a report of a pediatric patient with Williams syndrome presenting for inguinal hernia repair under general anesthesia.

CASE REPORT

A 5-month-old male infant, weighing 4.7 kg with Williams syndrome was scheduled for inguinal hernia repair under general anesthesia. The diagnosis of Williams syndrome was made at 4 months of age. Williams syndrome presented to cardiology clinic for evaluation of a systolic murmur. Evaluation by echocardiography in the cardiology clinic revealed mild supravalvular aortic stenosis with pulmonary artery stenosis. Preoperative evaluation revealed a small 5-month-old infant in no acute distress.. No cardiovascular, respiratory, digestive and renal systems abnormalities were detected. His vital signs were stable. Preoperative laboratory

evaluation including electrolytes, blood urea nitrogen, creatinine, calcium, and thyroid function tests were within normal limits. The hemoglobin was 12 g.dl-1. On the operation day the patient was accepted to the operating room. He was monitored with peripheral oxygen saturation (SpO₂), electrocardiogram (leads II, V1), cutaneous temperature (T), noninvasive blood pressure (NIBP), and endtidal carbon dioxide. Heart rate: 127 min⁻¹, blood pressure: 89/35 mmHg, body temperature 37.0 °C. Induction of anesthesia was done with sevoflurane and mixture 50% of O₂/N₂O, initiated sevoflurane with 1% until 8%. After completed induction and venous puncture was performed with a 26 G catheter in the left upper limb, and the trachea was intubated without difficulty with a 4.0 mm ID uncuffed tube. Maintenance was achieved using sevoflurane 1 MAC in a 2: 1 nitrous oxide : oxygen mixture and intravenous fentanyl 1,5 mcg.kg⁻¹.h⁻¹. Hemodynamic and other vital parameters were stable during intraoperative period. The duration of the surgery was 40 minutes. After recovery of muscle tone, spontaneous breathing was adequate, trachea was extubated. No respiratory or hemodynamic problems were occurred. The patient was admitted to recovery room with stable vital signs (blood pressure: 88/39 mmHg, heart rate 126 beats.min⁻¹, peripheral oxygen saturation 99%, body temperature 37.0 °C). Thirty minutes later he was sent to service with stable vital signs.

DISCUSSION

The multisystem involvement associated with Williams syndrome raises many considerations for the anesthetic management of these patients. Of primary concern is the cardiovascular pathology including congenital heart defects such as SVAS. At least 22 cases of sudden death in patients with Williams syndrome have been documented in the literature. The mechanism for sudden death in these patients includes myocardial ischemia, decreased cardiac output, and ventricular arrhythmias (4,5,6). Thus, a thorough cardiac evaluation for all patients with Williams syndrome prior to anesthesia and surgery is necessary to identify high-risk patients. Horowitz et al. (7) suggest that the preoperative work up should also include coronary angiography for all patients with Williams syndrome requiring anesthetic care. These patients may present with difficult mask ventilation and tracheal intubation during induction of anesthesia because of a flattened midface, wide mouth, and dental malocclusion (8). We performed mask ventilation and tracheal intubation without problem. Nearly all patients with Williams syndrome have some degree of musculoskeletal involvement, including joint laxity that may lead to muscular weakness. Lipid storage in muscles and increased variability in fiber size has also been noted (1,6). This may be an anesthetic concern given that patients with other conditions with similar biopsy findings have variable reactions to neuromuscular blockade including prolonged neuromuscular blockade in patients with Duchenne's muscular dystrophy after administration of nondepolarizing neuromuscular blocking agents (8,9). No muscle relaxant was used during the operation in our case.

Other features, such as the relationship of Williams syndrome with neonatal hypercalcemia, altered neurodevelopment, and hypothyroidism have not been shown to be directly related to the deletion of elastin, thus additional genetic abnormalities may yet be described. Several cases have been reported of patients with elevated TSH levels and normal FT4 levels (10). Preoperative

laboratory evaluation including electrolytes, blood urea nitrogen, creatinine, calcium, and thyroid function tests were within normal limits in our patient.

Anesthesia can be safely administered to children with Williams syndrome. We think that general anesthesia can be performed successfully with hemodynamic stability.

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References

1. Lashkari A, Smith A, Graham J. Williams - Beuren Syndrome. *Clin Pediatr* 1999;38:189-208.
2. Williams JC, Barratt-Boyes BG, Lowe JB. Supraventricular aortic stenosis. *Circulation* 1961;24:1311-1318.
3. Ewart AK, Morris CA, Atkinson DL et al. Hemizygoty at the elastin locus in a developmental disorder, Williams syndrome. *Nat Genet* 1993;5:11-16.
4. Bird LM, Billman GF, Lacro RV et al. Sudden death in Williams syndrome: report of 10 cases. *J Pediatr* 1996;129:926-931.
5. Van Son JAM, Edwards WD, Danielson GK. Pathology of coronary arteries, myocardium, and great arteries in supravalvular aortic stenosis. *J Thorac Cardiovasc Surg* 1994;108:21-28.
6. John Medley, Pierantonio Russo, Joseph D. Tobias. Perioperative care of the patient with Williams syndrome. *Pediatric Anesthesia* 2005;15:243-247.
7. Horowitz PE, Akhtar S, Wulff JA et al. Coronary artery disease and anesthesia related death in children with Williams syndrome. *J Cardiothorac Vasc Anesth* 2002;6:739-741.
8. Tobias JD, Atwood R. Mivacurium in children with Duchenne's muscular dystrophy. *Pediatr Anesth* 1994;4:57-60.
9. Frankowski GA, Johnson JO, Tobias JD. Rapacuronium administration to two children with Duchenne's muscular dystrophy. *Anesth Analg* 2000;91:27-28.
10. Cammareri V, Vignati G, Gianfranco N et al. Thyroid hemiagenesis and elevated thyrotrypsin levels in a child with Williams syndrome. *Am J Med Genet* 1999;85:491-494.

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