

## Anaesthetic management of Williams syndrome

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Williams syndrome is a rare, genetic, multisystem disorder warranting significant anaesthetic concerns. Sudden cardiac death has been reported in the perioperative period and attributed to cardiovascular pathology. Present case report highlights the risk and meticulous management for safe perioperative outcome of such patients.

**Keywords:** Williams syndrome; aortic stenosis; anaesthesia

### Introduction

Williams syndrome is caused by hemizygous microdeletion on chromosome 7; the estimated prevalence of 1:20,000 live births. The syndrome was first described by Williams, Barratt-Boyes and Lowe in 1961. It is characterized by cardiovascular disease, distinctive facies and personality, hypercalcaemia, and connective tissue growth and endocrine abnormalities.<sup>1</sup> Sudden cardiac death has been reported in the perioperative period and attributed to cardiovascular pathology.<sup>2-4</sup> Here we are reporting a case of successful perioperative management of a paediatric patient who underwent a laparotomy for strangulated inguinal hernia.

### Case report

An 8 years old male child weighing 18kgs was referred to our tertiary care centre for emergency laparotomy for strangulated inguinal hernia. He was diagnosed to be a case of Williams syndrome at the age of 1 year. History suggestive of failure to thrive and delayed milestones were present. Physical examination revealed characteristic dysmorphic features, dental malocclusion and grade III systolic murmur all over precordium, radiating to neck and back. Heart rate and blood pressure were 112/min and 90/50mmHg respectively with normal oxygen saturation. Investigations showed haemoglobin 11gm%, leucocytosis and borderline serum calcium levels. Echocardiogram was suggestive of biventricular outflow tract obstruction and severe pulmonary hypertension. Fasting status was confirmed. Potential risks of anaesthesia and surgery were explained to the parents; written, informed consent was obtained.

Child was premedicated with intranasal dexmedetomidine 1µ/kg using an atomizer device 20mins prior to induction, in the preoperative holding area to provide anxiolysis and facilitate parental separation. Once a satisfactory sedation score was achieved, child was shifted to operating room. A wide bore intravenous cannula was secured and antibiotic prophylaxis was given. Standard monitors: electrocardiogram, pulse oximetry, invasive blood pressure, temperature and capnogram were applied. After preoxygenation, anaesthesia induction was performed by intravenous thiopentone 5mg/kg, fentanyl 2µ/kg and atracurium 0.5mg/kg. Airway was secured with 5.5 uncuffed endotracheal tube. During laryngoscopy, visualization of vocal cords was difficult and required external laryngeal manipulation.

Anaesthesia was maintained using 0.8 – 1 MAC of sevoflurane in oxygen-air mixture. Intraoperative tachycardia during incision and abdominal exploration was managed by small boluses of esmolol. Blood pressure measurements were stable throughout the procedure. Surgery lasted 90mins and adequate haemostasis was confirmed. Blood loss of 200ml was replaced by crystalloids. At the end of surgery, bilateral transverse abdominis plane block was performed using 20ml volume of 0.125% bupivacaine. After regaining of protective airway reflexes, patient was extubated awake and shifted to ICU for further management. Postoperative period was uneventful. Pain relief lasted for 18hrs. Antiemetics and rectal paracetamol



suppositories were prescribed for the rest of inpatient course.

### Discussion

Williams syndrome is a rare, complex, multisystem disorder of genetic pathology; poses significant perioperative risk to anaesthesiologists. Sudden death has been reported as a very common complication of anaesthesia, surgery and other procedures in these patients.<sup>4,6</sup> Structural cardiovascular abnormalities occur in 80% of all patients and are present in up to 93% of these patients presenting in the first year of life. Reported cardiac lesions include supra-aortic stenosis(75%), pulmonary artery stenosis(37%), coronary artery abnormalities(9%) and other systemic arteriopathies. The risk of sudden cardiac death is 25 to 100 times than that in the general population. The reason for this is not completely understood. The risk of sudden cardiovascular collapse appears to be greater in the presence of bilateral outflow tract obstruction, especially with coronary artery stenosis. Prolongation of corrected QT interval on ECG contribute to the increased risk of sudden death.<sup>5</sup>

To date, no studies have evaluated anaesthetic regimens in patients with Williams syndrome, and no evidence based recommendations are available. As these patients exhibit distinctive behavioural and emotional traits, preoperative anxiolysis and sedation is mandatory to prevent physiological and psychological stress associated with the disease. Intranasal dexmedetomidine was used as it produces effective sedation for parental separation.<sup>7</sup>

Thiopentone provided smooth and safe anaesthesia induction in our patient. Use of inhalational agents as sole agents is debatable as the resulting vasodilation produce decreased venous return, cardiac output and compromised myocardial perfusion in the settings of aortic stenosis and coronary arteriopathy.

Dysmorphic features in these patients include short upturned nose, flat nasal bridge, long philtrum, wide mouth, flat malar areas, full lips, dental malocclusion and widely spaced teeth, micrognathia, and periorbital fullness.<sup>5</sup> These patients may present difficult mask ventilation and tracheal intubation during induction. We performed ventilation and intubation without

many problems. External laryngeal manipulation was needed for visualization of cords. Medley et al reported concerns regarding their patient that trachea was intubated with a smaller endotracheal tube than expected for age and developed postoperative stridor because of laryngeal oedema.<sup>3</sup> Invariably these patients have some degree of musculoskeletal involvement, including joint laxity and muscle weakness. Titration of dosage of non-depolarizing neuromuscular blocking agents to train-of-four responses is preferred. Risk of masseter spasm and malignant hyperthermia must be considered if succinyl choline and other triggering agents are used during induction.<sup>8</sup>

Precise defect of hormonal control remains uncertain; disturbances of calcium homeostasis<sup>1</sup> and hypothyroidism are common features.<sup>9</sup> Urologic abnormalities, both structural and functional, are seen with increased frequency in Williams syndrome. Nephrocalcinosis occurs secondary to hypercalcaemia and renal function may decline with age.<sup>10</sup> Hence, we suggest thorough work up on thyroid function and calcium balance is essential to avoid medical complications during perioperative period.

A detailed preoperative assessment including cardiac evaluation, airway anatomy, metabolic function and cognitive status plays a crucial role for planning and executing safe anaesthetic interventions regardless of surgical procedure. Intensive monitoring during intra and postoperative periods forms basis for successful anaesthetic outcome of these patients.

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