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Case Report

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Anesthetic Management of a Patient With Trisomy 18 Undergoing a Multilevel Spinal Fusion

Davis Frease[a, c], David Rico Mora[b]

Abstract

Trisomy 18 is the second most common autosomal trisomy aside from trisomy 21. Anesthesiologists were unlikely to manage such patients in the past, specifically those surviving later into childhood due to the 90% mortality rate within the first year of life and the lack of procedural options that were available. However, a paucity of literature regarding the anesthetic management of such patients exists. Trisomy 18 patients present a unique anesthetic challenge, given the presence of associated dysmorphic facial features and the involvement of multiple organ systems, leading to difficult airway and hemodynamic disturbances. In this case report, we present the anesthetic management of a 9-year-old patient with trisomy 18 undergoing a multilevel spinal fusion. Despite significant intraoperative hemorrhage, the patient was able to tolerate the procedure without complications, likely owing to the meticulous preoperative preparation and the patient's survival later into childhood. This case contributes to a small subset of literature which suggests that patients with trisomy 18 who survive later into childhood have an improved ability to tolerate general anesthesia.

Keywords: Trisomy 18 perioperative management; Pediatric multilevel spinal fusion; Pediatric intraoperative hemorrhage; Pediatric intraoperative hypovolemic shock

Introduction

Trisomy 18, also known as Edward's syndrome, is the second most common autosomal trisomy aside from trisomy 21. Features of trisomy 18 include: dysmorphic facial features and malformations of the cardiac, respiratory, gastrointestinal, genitourinary and central nervous systems [1]. These features present a unique challenge to the anesthesiologist given the higher prevalence of difficult intubation, ventilation, oxygenation, and complex hemodynamic management associated with underlying cardiac pathology.

In this case report, we describe the anesthetic management of a 9-year-old patient with trisomy 18 and neuromuscular scoliosis undergoing a T2 - S1 spinal fusion.

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[a]Department of Anesthesiology, Walter Reed National Military Medical Center, Bethesda, MD, USA
[b]Department of Anesthesiology, Pain and Perioperative Medicine, Children's National Hospital, Washington, DC, USA
[c]Corresponding Author: Davis Frease, Department of Anesthesiology, Walter Reed National Military Medical Center, Bethesda, MD, USA.
Email: davis.frease@gmail.com

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Case Report

Investigations

The patient was a 9-year-old female with complete trisomy 18 and neuromuscular scoliosis presenting for a T2 - S1 spinal fusion. The patient was gastric-tube (G-tube) dependent and had a history of severe acid-reflux resulting in occasional pulmonary complications. Baseline pulmonary function was not impacted by her scoliotic spine. Nevertheless, formal pulmonary function testing was not obtained. The preoperative echocardiogram showed bicuspid aortic valve, mild aortic valve regurgitation, and normal ventricular function. No atrial or ventricular septal defects were identified on the echocardiogram. Arrhythmia history was negative. Preoperative cardiology clearance was requested prior to surgical intervention. Preoperative radiographs of the thoracic spine demonstrated a Cobb angle of 70° in neutral view. Lateral flexion views demonstrated 93° levoconvex curvature with right sided bending and 52° levoconvex curvature with left sided bending.

Approximately 3 months prior to the scheduled procedure, the patient was hospitalized for acute on chronic malnutrition. Prior to discharge, she was evaluated by the preoperative care clinic, which recommended ongoing optimization of her nutritional status, oral iron treatment and initiation of a pulmonary sick plan 3 days prior to the procedure. The patient weighed 16.7 kg. The airway examination was remarkable for micrognathia, dysmorphic facial features and limited neck extension. Laboratory analysis from her hospitalization was notable for normal coagulation studies and a normal liver panel, including a normal albumin level.

Diagnosis and treatment

Preoperatively, methadone (1.5 mg) and aprepitant (15 mg) were administered through the patient's G-tube. The operating

room (OR) was pre-staged with ultrasound, difficult airway equipment and one unit of packed red blood cells (pRBCs) was available prior to surgical incision. Standard noninvasive monitoring was used to monitor the patient's vital signs and somatosensory evoked potentials (SSEPs) and motor evoked potentials (MEPs) were utilized throughout the case for neuromonitoring. Autologous blood salvage (cell saver) was available during the procedure.

Sevoflurane mask induction was started, and initial ultrasound-guided intravenous (IV) access was obtained in the left antecubital vein afterward. Subsequently, 80 mg of IV propofol was administered to facilitate endotracheal intubation, manual ventilation was adequate and endotracheal intubation was easily achieved via direct laryngoscopy. A right radial arterial line and additional peripheral IV access was obtained in the right wrist and left saphenous vein following intubation respectively. Anesthesia maintenance was achieved with a remifentanil infusion and sevoflurane (0.5 minimal alveolar concentration) with frequent titration in response to hemodynamic derangements. Tranexamic acid bolus followed by an infusion was administered to decrease bleeding risk.

The initial point of care laboratory analysis was normal. Refractory hypotension started approximately 3 h following the initial surgical incision. At first, hypotension was unresponsive to decreases in anesthetic concentrations. Significant bleeding was noted by the surgical team and a second point of care laboratory analysis showed a decrease in the patient's hemoglobin to 8.6 mg/dL (baseline hemoglobin 10.8 mg/dL) and a base excess of -3.2. Subsequently, transfusion of pRBCs was started given the ongoing hemorrhage, significant hypotension, and hemodynamic instability. Cell salvage was utilized during the case and autologous blood was transfused near the conclusion of the case given persistent hypotension. While awaiting cell salvage blood, 2.5 mg bolus of IV ephedrine was administered to treat the hypotension.

In total, 333 mL of pRBCs and 180 mL of autologous red blood cells were transfused. Hypotension was responsive to volume resuscitation with both red blood cells and crystalloids (1.7 L of Ringer's lactate solution). The estimated blood loss was 650 mL, and urine output was 320 mL. The experienced blood loss represented 52% of the patient's estimated blood volume and the anesthetic duration was 7.5 h approximately.

Follow-up and outcomes

The patient was extubated at the conclusion of the case and transferred to the pediatric intensive care unit (PICU) on blow-by oxygen. Pain was well controlled postoperatively with scheduled diazepam, oxycodone, ketorolac, acetaminophen, and her breakthrough pain was treated with IV morphine as needed. The patient was discharged home on postoperative day 7.

Discussion

As there are a limited number of case reports in the anesthesia literature of patients with trisomy 18, no definitive protocol exists for their perioperative management, specifically for those patients surviving late into childhood. Nowadays, there is a significantly higher number of anesthesiologists managing patients with trisomy 18 due to the development of newer surgical interventions aimed to treat these patients. Thus, further contribution to the literature regarding their perioperative care will improve the available data to achieve the safest and most effective medical management.

Patients with trisomy 18 presenting for surgical procedures are likely to have malformations of multiple organ systems, which may make their anesthetic management challenging. Structural heart defects occur in nearly 90% of patients, most commonly septal defects, patent ductus arteriosus, and polyvalvular disease. Gastrointestinal malformations in such patients include: omphalocele, esophageal atresia with tracheoesophageal fistula, pyloric stenosis, and Meckel's diverticulum. Poor feeding frequently results in G-tube placement and malnutrition. A high prevalence of gastroesophageal reflux disease predisposes these patients to recurrent pneumonia secondary to aspiration. Other pulmonary problems such as upper airway obstruction, central apnea, and pulmonary hypertension may also be prevalent [2].

A recent large cohort review encompassing 11 patients at a US tertiary care center concluded that over the span of 6 years, nine patients required anesthesia for a total of 121 encounters. Less invasive procedural interventions include the following: diagnostic and surgical airway procedures, feeding tube placements and revisions, ear examinations and tube placements, peripheral venous catheter placements, esophagogastroduodenoscopy and/or colonoscopy. Higher risk procedures include scoliosis repair surgery and cardiac surgery. Notably, most observed procedures were performed during infancy [3].

Previously, cardiac defects in trisomy 18 patients were managed conservatively, as they were not thought to impact survival. However, the surgical correction of cardiac defects has become increasingly popular in trisomy 18 patients after recent studies have shown an 82-91% survival rate in patients undergoing operative management. Trisomy 18 patients that do not undergo surgical correction of cardiac defects have a higher prevalence of heart failure, pulmonary hypertension, and death [2]. The increased risk for development of neoplasia in those with trisomy 18 also offers the potential for surgical intervention. Hepatoblastoma resection has been reported in three patients, two of which survived without recurrence in the following 2 years [2].

Scoliosis is a common finding in older children with trisomy 18, as was seen in our patient. Cereda et al urged correction in those with severe scoliosis given the potential risk of restrictive lung disease [2]. The Support Organization of Trisomy 18, 13 and Related Disorders (SOFT), which maintains a registry of surgical procedures performed in children with trisomy 18, indicated that 34 spinal fusion procedures have been performed in those without mosaicism.

The current literature illustrates the rising trend towards procedural management in patients with trisomy 18 and the resultant increased need for anesthesia services. While several case reports exist describing the anesthetic management of patients with trisomy 18 younger than 5 years of age, few have

described in detail the anesthetic management of a patient with trisomy 18 in late childhood, likely related to the 90-95% mortality rate during the first year of life [2], and a survival to age 6 previously reported to be as low as 3% [4]. The first report of an older child with trisomy 18 receiving general anesthesia and caudal epidural anesthesia for heel cord lengthening was characterized by an uneventful intraoperative course [5]. Nevertheless, the large cohort review encompassing 11 patients with trisomy 18 did include two patients, who were 16 and 11 years of age, respectively; combined, these two patients received a total of 31 anesthetics with complications related to difficult intubation, laryngospasm, and bradycardia. Other anesthetic complications cited in the review included difficult bag-valve-mask ventilation, difficult IV access and cardiac events necessitating resuscitation, which included cardiopulmonary reanimation (CPR) in younger patients [3]. Further, a recent retrospective review of airway management of trisomy 18 patients found that there was a decreased risk of difficult intubation in patients older than 6 years of age compared with their younger counterparts [6].

In this case, a 9-year-old patient with trisomy 18 was able to tolerate a major orthopedic procedure with significant intraoperative hemorrhage, representing a total loss of nearly 50% of her estimated blood volume. Multi-level spinal fusion infers an increased risk of intraoperative hemorrhage in the general population. However, in patients with trisomy 18, this risk may be further complicated by the presence of cardiac lesions which may increase the possibility of perioperative complications.

Meticulous preoperative preparation for potential intraoperative complications allowed for rapid diagnosis and successful management of complications, specifically intraoperative hemorrhage. In our case, this included establishing adequate IV access for volume resuscitation, placement of an arterial line for rapid recognition of hypotension, prestaging the OR with laboratory analysis capabilities, having pRBCs available in the OR prior to case start, and utilizing cell saver capabilities for the case. Given the significant intraoperative blood loss associated with the surgical intervention, prestaging the OR with an additional unit of pRBCs may have been warranted.

While this case suggests that patients with trisomy 18 surviving later into childhood may tolerate major surgical intervention without significant morbidity, the absence of significant comorbidities and genetic heterogeneity could make generalization of this case to a broader cohort difficult. Courreges et al described the anesthetic management of a 7-year-old patient with trisomy 18 and major cardiovascular disease including pulmonary hypertension, who tolerated general anesthesia without complications while undergoing a Cohen procedure [7]. Further, Tsukamoto et al described the anesthetic management of a 12-year-old patient with trisomy 18 and untreated tetralogy of Fallot, who tolerated general anesthesia undergoing dental rehabilitation [8]. Thus, survival into later childhood may be a predictor of ability to tolerate general anesthesia. However, unlike these two cases, our patient did not have major cardiac disease; her ability to tolerate major hemodynamic changes associated with intraoperative hemorrhage presumably was significantly impacted by this fact.

Adequate preoperative evaluation and optimization of underlying pathologies in trisomy 18 patients are paramount to minimize intraoperative complications, specifically those with complex cardiac and pulmonary pathologies. Given the propensity for dysmorphic features, anticipating the presence of difficult mask ventilation and intubation should be the standard of care. It is also important to highlight that the obtention of vascular access in patients with trisomy 18 could be challenging and the use of ultrasound could improve the success rate, decreasing the number of failed attempts.

Conclusions

In conclusion, we were able to successfully manage a multilevel spinal fusion with intraoperative hemorrhage in a 9-year-old patient with trisomy 18. This case seeks to contribute to the limited available literature regarding the anesthetic management of patients with trisomy 18, specifically in those surviving later into childhood, in the face of an increasing trend towards more aggressive procedural care in such patients. Other case reports and recent reviews have indicated that there may be a correlation with trisomy 18 patients' survival later into childhood and an increased ability to tolerate general anesthesia with decreased risk for intraoperative complications. This case adds further evidence to such a correlation. Seemingly this is secondary to the absence of significant organ malformations which could result in significant pathologic states in such patients surviving later into childhood.

Learning points

The current literature describing trisomy 18 patients surviving later in childhood offers an indication that such patients are perhaps more likely to tolerate major surgical intervention and associated anesthesia than their younger counterparts. Nevertheless, ample preoperative preparation for a potentially difficult airway, difficult vascular access, and challenging hemodynamic management is paramount in such patients given the clinical features associated with trisomy 18. As the breadth of available literature expands and the increasing trend towards procedural intervention evolves, more definitive conclusions regarding their anesthetic management will likely be elucidated in the future.

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None to declare.

Conflict of Interest

None to declare.

Informed Consent

Not applicable

Author Contributions

DF and DRM provided care for the patient. DF performed initial case review and manuscript preparation, literature review, and editing of subsequent revisions. DRM was involved in the literature review, initial draft, subsequent revisions, and editing of the manuscript.

Data Availability

The data supporting the findings of this study are available from the corresponding author upon reasonable request.

Abbreviations

G-tube: gastric tube; OR: operating room; pRBCs: packed red blood cells; SSEPs: somatosensory evoked potentials; MEPs: motor evoked potentials; IV: intravenous; PICU: pediatric intensive care unit

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