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Case Report

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Anesthetic Implications in Rubinstein-Taybi Syndrome

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Abstract

Rubinstein-Taybi syndrome (RTS), also known as broad thumb-hallux syndrome or Rubinstein syndrome, is an autosomal dominant condition characterized by short stature, moderate to severe learning difficulties, distinctive facial features, and broad thumbs and first toes. Given the associated anatomical features of the disorder, anesthetic care may be required during surgical procedures to correct the end-organ effects of the disorder. We present a 5-year-old boy with RTS that required anesthetic care for an adenotonsillectomy. Previous reports of anesthetic care for patients with RTS are reviewed and the perioperative considerations of these patients are discussed.

Keywords: Rubenstein-Taybi syndrome; Anesthetic; Airway

Introduction

Although the constellation of features in Rubinstein-Taybi syndrome (RTS) was first reported in the French literature in 1957, a larger series was subsequently reported by Jack Herbert Rubinstein and Hooshang Taybi in 1963 [1, 2]. RTS is a rare disorder characterized by high arched palate, broad thumbs, prominent forehead, high-arched eyebrows, antimongoloid slant of the palpebral fissures, beaked nose with flat bridge, abnormal ears, and mental retardation [1-3]. It may be inherited as an autosomal dominant disorder or arise as a spontaneous mutation. Other associated phenotypic features include microcrania, short stature, ocular defects, deviated nasal septum, incompletely descended testes, finger and toe abnormalities, hyperextensible joints, and hyperactive patellar reflexes. Patients frequently have associated respiratory and feeding difficulties as well as associated congenital heart disease [4]. The prevalence of RTS is not precisely known, but the incidence is estimated to be approximately 1 in 300,000 in the general population [3]. Discovered 30 years after the initial clinical reports, RTS results from submicroscopic 16p13.3 deletion of the response element-binding protein (CREBBP) gene [5]. The CREBBP gene controls production of a protein that regulates the activity of several other genes and therefore plays an important role in regulating cell growth and division which is essential for normal fetal development. We present a 5-year-old boy with RTS that required anesthetic care for an adenotonsillectomy. Previous reports of perioperative care of patients with RTS are reviewed and options for anesthetic care are presented.

Case Report

Institutional Review Board approval is not required at Nationwide Children's Hospital (Columbus, OH) for the presentation of single case reports. The patient was a 5-year-old, 18.8 kg boy who presented for adenotonsillectomy for sleep-disordered breathing. His past medical history was significant for RTS diagnosed at 5 years of age, sepsis, plagiocephaly, thrombocytopenia, neutropenia, atrial septal defect (ASD), history of tachycardia during the newborn, anemia, unilateral inguinal hernia, methicillin-resistant *Staphylococcus aureus* (MRSA) colonization, and retinopathy of prematurity (ROP). His past surgical history included a flexible esophagogastroduodenoscopy (EGD) with direct placement of a percutaneous G-tube, upper GI endoscopy, G-J tube removal, circumcision, exploration of undescended testicle, tympanostomy tube placement, and previous adenoidectomy. Associated co-morbid conditions included attention-deficit disorder, scoliosis, lack of normal physiological development, developmental speech or language delay, sleep-disordered breathing, and allergic rhinitis. Current home medications included dextroamphetamine-amphetamine extended release (10 mg by mouth once daily in the morning), polyethylene glycol (8.5 g by mouth once daily, adjusted to give 1 - 2 soft stools daily), daily iron supplement, ibuprofen (150 mg oral suspension by mouth once daily), loratadine (5 mg by mouth once daily), and a multivitamin (1 tab by mouth

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once daily). Allergies included only seasonal problems. Preoperative physical examination revealed a child in no acute distress with moderate scoliosis, dysmorphic facies, broad toes, and tonsillar hypertrophy. Airway examination revealed a Mallampati grade II view. Perioperative laboratory evaluations including electrolytes, urinalysis, coagulation function, glucose, and hepatic function were normal. The last hemoglobin was 13.4 g/dL. He had a sip of water with his morning medications, but otherwise was nil per os for 6 h. The preoperative room air oxygen saturation was 100%. The patient was transported to the operating room and routine American Society of Anesthesiologists' monitors were applied. Anesthesia was induced with the inhalation of sevoflurane in a mixture of nitrous oxide. A 22-gauge peripheral intravenous cannula was placed following anesthetic induction. Bag-valve-mask ventilation was provided without difficulty. Propofol (2 mg/kg) and morphine (0.05 mg/kg) were administered intravenously. Direct laryngoscopy was performed with a Miller 2 blade, which revealed a Cormack-Lehane grade I view. A 4.0 mm cuffed endotracheal tube was placed on the first attempt and a small amount of air added to the cuff to seal the airway at 20 - 25 cmH_2O. The patient was positioned supine with a shoulder roll in place and arms tucked to the side. No intraoperative antibiotics were given. The heart rate varied from 130 to 170 beats per minute with a normal sinus rhythm. No bradycardia or arrhythmias were noted. Intraoperative fluids included 250 mL of lactated Ringers solution. Dexamethasone (4 mg) and ondansetron (2 mg) were administered intravenously to provide postoperative inflammation and nausea relief, respectively. Following completion of the surgical procedure, the patient was transferred to the post-anesthesia care unit (PACU) for observation of hemodynamic and respiratory function. Per our usual routine, his trachea was extubated in the PACU when he was awake. Postoperative pain control was provided with hydrocodone-acetaminophen every 6 h as needed. He was admitted to the inpatient ward for 8 h of postoperative observation. The remainder of his postoperative course was uncomplicated and he was discharged home the same day.

Discussion

As with the anesthetic care of all patients, the focus of effective perioperative care of patients with RTS begins with the preoperative examination and the identification of end-organ involvement by the primary disease process. Of primary concern to anesthesia providers are problems with airway management which have been previously noted in several anecdotal case reports from the literature [6-8]. Difficulties with direct laryngoscopy and endotracheal intubation are primarily related to specific craniofacial anatomical features including high-arched palate, hypoplasia of the mandible (micrognathia), and limited mouth opening. Although we were able to successfully accomplish endotracheal intubation using direct laryngoscopy without significant difficulties, the ability to accomplish adequate bag-valve-mask ventilation should be demonstrated prior to the use of neuromuscular blocking agents. Additionally, the appropriate equipment for dealing with the difficult airway should be readily available including indirect laryngoscopy tools [9, 10]. Anecdotal success has been reported with the use of a laryngeal mask airway for elective airway management or as a conduit for endotracheal intubation following failed routine airway management [6, 11]. The risk-benefit of such a practice must be weighed given high incidence of gastroesophageal reflux in RTS patients. In our patient, inhalational induction with sevoflurane was performed as the most humane way to achieve intravenous access followed by endotracheal intubation with a cuffed endotracheal tube.

Perioperative respiratory and ventilation issues may be further complicated by congenital tracheal stenosis, abnormal pulmonary lobulation, excessive mucous secretion, and gastroesophageal reflux [7, 12, 13]. The potential co-morbid condition of the respiratory system is illustrated by the fact that respiratory infections and congenital heart disease are the leading causes of death during the first year of life. Copious secretions have been noted by other authors which may make inhalation induction problematic or complicate the immediate postoperative period [6]. The perioperative respiratory problems may be further magnified in patients with preoperative symptoms of sleep-disordered breathing as were noted in our patient [14, 15]. Whenever feasible, short-acting anesthetic agents are preferable to limit their effect on postoperative respiratory function. The use of regional has been suggested as a means of limiting perioperative opioid needs thereby limiting their effects on respiratory function [16]. Considering the potential co-morbid involvement of the airway and respiratory system, postoperative monitoring is suggested with continuous pulse oximetry or in an ICU setting for more complex surgical procedures.

Associated congenital heart disease occurs in up to 25-30% of patients with RTS [17, 18]. Reported defects have included atrial septal defect, ventricular septal defect, patent ductus arteriosus, coarctation of the aorta, pulmonic stenosis, bicuspid aortic valve, pseudotruncus, aortic stenosis, vascular rings, and hypoplastic left heart syndrome. In addition to structural cardiac defects, conduction disturbances have been occasionally reported, many in association with the administration of perioperative medications including succinylcholine or atropine and neostigmine for reversal of neuromuscular blockade [19, 20]. Stirt reported short runs of supraventricular tachycardia and multifocal premature ventricular and atrial contractions for 5 min following the administration of succinylcholine [20]. No hemodynamic changes were noted, no treatment was required, and the case proceeded uneventful. Similar problems were noted in the same patient during a prior anesthetic with the administration of atropine and neostigmine. Given the potential for such involvement, preoperative echocardiography and electrocardiography should be obtained based on the clinical examination and history.

Although anecdotal, the use of succinylcholine should generally be avoided. When neuromuscular is required, non-depolarizing agents are suggested. Given the potential pro-arrhythmogenic effects of medications with vagolytic effects, there may be a theoretical advantage to the use of cis-atracurium over rocuronium for non-emergent airway management [21]. Alternatively, endotracheal intubation can be accomplished without neuromuscular blockade as noted in our pa-

tient with the combination of sevoflurane and propofol. For intravenous induction, others have reported acceptable conditions for endotracheal intubation using a combination of propofol and remifentanil [22, 23].

In summary, we present concerns regarding the anesthetic management of a patient with RTS. Of primary concern are anecdotal reports regarding anatomical features of the syndrome which may make direct laryngoscopy and endotracheal intubation problematic. Associated respiratory disabilities may lead not only to intraoperative, but also postoperative respiratory insufficiency. Potential co-morbid involvement of the cardiac system includes congenital heart disease and the potential for conduction disturbances.

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