

Anesthetic Management of an Asian Pediatric Patient With Rubinstein-Taybi Syndrome for Dental Surgery

Phui Sze Au Yong^{a, c}, Hua Ling Evangeline Lim^b

Abstract

Rubinstein-Taybi syndrome (RTS) is a rare genetic disorder that is associated with dysmorphism, moderate to severe intellectual disability and abnormalities involving multiple organ systems. For anesthetists, these patients pose unique challenges in the administration of anesthesia and airway management. We describe the anesthetic conduct of a pediatric patient with RTS coming for dental surgery as a day surgery case and will discuss the anesthetic considerations for such patients.

Keywords: Rubinstein-Taybi syndrome; Pediatrics; Case report; Difficult airway; Anesthesia; Dental surgery

Introduction

Rubinstein-Taybi syndrome (RTS) is a well-defined complex of congenital malformations characterized by facial abnormalities, broad thumbs, big toes and intellectual disability. It is a rare disorder first described in 1963 [1]. It can be inherited as an autosomal dominant trait or as a spontaneous mutation in approximately 25% of the patients, involving submicroscopic interstitial deletions within 16p13.3 of the CREB-binding protein gene (CREBBP) [2]. This protein regulates other genes involved in cell growth and division which is essential for normal fetal development. Mutations in the EP300 gene have also been identified in 3-8% of individuals. Diagnosis of this condition is primarily through recognition of physical features such as down slanted palpebral fissures, low hanging columella and/or broad nose bridge, high-arched palate, cusp-like structures

on the front teeth, and large and/or angulated thumbs and toes. The diagnosis may be further supported by genetic tests and characteristic features on X-rays of the hands and feet. Multiple organ systems may be involved including the cardiovascular, respiratory, gastrointestinal and urological systems. These individuals are also at higher risk of malignancies such as lymphoma or leukemia.

The literature on anesthetic management of RTS patients are mostly single case reports or case series, reflecting the rarity of the disease. Many of the early publications are from Europe [3-5], with later publications appearing from South Africa [6], India [7], Middle East [8] and Korea [9]. The worldwide prevalence is estimated to be about 1 in 100,000 to 1,250,004. Stevens et al reviewed 50 such patients and noted frequent hospitalizations and operations, finding that each patient may undergo an average of 2.7 anesthetics [10]. There were anesthetic problems in nine cases, including respiratory distress, apnea and prolonged recovery.

Anesthetic challenges occur mainly due to craniofacial, cardiac and skeletal anomalies [7, 9, 11]. Skeletal anomalies that could have implications for anesthetists include kyphoscoliosis, and abnormalities of the vertebrae and pelvis with associated spinal cord tethering. Approximately one-third of children with RTS have congenital heart disease such as ventricular or atrial septal defects and/or conduction anomalies which predispose them to intraoperative hypotension and arrhythmias. Suxamethonium [12], neostigmine and atropine appear to cause arrhythmias more frequently in patients with RTS than those without.

Children with RTS are known to have potentially difficult airways with a tendency to obstruct. They are also recognized to be at higher risk of aspiration. Congenital tracheal stenosis, tracheomalacia, abnormal pulmonary lobation have been described in these children and these have implications for ventilation. Difficult laryngoscopy and intubation should be anticipated especially in the presence of micrognathia and macrostomia. There is a high incidence of obstructive sleep apnea, which may cause hypoxia during recovery from anesthesia or sedation [13]. All these predispose them to perioperative respiratory complications.

Individuals with RTS have intellectual disability with intelligence quotient (IQ) ranging from 36 to 51. There might be other central nervous system disorders such as hypotonia, seizures, hyper-reflexia, hearing impairment, speech and other developmental delays. In addition, there might be behavioral issues such as sensory intolerance with noise and crowds,

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^aDivision of Anaesthesiology and Perioperative Medicine, Singapore General Hospital, Singapore 169608, Singapore

^bDepartment of Paediatric Anaesthesia, KK Women's and Children's Hospital, Singapore 229899, Singapore

^cCorresponding Author: Phui Sze Au Yong, Department of Surgical Intensive Care, Division of Anaesthesiology and Perioperative Medicine, Singapore General Hospital, Singapore 169608, Singapore.
Email: angie.au.yong.p.s@singhealth.com.sg

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short attention span, impulsiveness and moodiness. These may make communication, cooperation with anesthetic induction, pain assessment and management more challenging.

Craniofacial growth retardation in RTS is frequently complicated by unerupted teeth and dental caries which is common when dental hygiene is difficult in intellectually deficient children [14]. Dental surgery frequently requires nasal intubation. However, choanal atresia or deviated nasal septum may make nasal intubation in these children more difficult. Therefore, meticulous planning of airway management is needed. The objective of this case report is to highlight anesthetic considerations for pediatric patients with RTS who are listed for day surgery that involves airway manipulation.

Case Report

A 13-year-old, 40 kg, and 1.5 m tall Chinese boy with RTS was electively listed for restoration of severe dental caries at KK Women's and Children's Hospital, a specialist pediatric hospital in Singapore. Born full term, he had surgical correction of patent ductus arteriosus (PDA) during neonatal period but no other cardiac issues. He was diagnosed with RTS in the USA in the course of being worked up for intellectual disability through genetic testing. There was no family history of RTS.

Physical manifestations included craniofacial dysmorphism (prominent forehead, beaked nose with flat nose bridge, abnormal ears), global development delay and severe myopia. He was fretful, expressed only non-coherent sounds and wheelchair dependent. His father mentioned a previous traumatic experience of fitting him for reading glasses in which he required physical restraint by multiple adults. He was not on long-term medications. Although he had a small mouth and his Mallampati score could not be assessed, there was good mouth opening, a thyromental distance > 6 cm and normal neck extension. He had copious oral secretions but no gastric reflux or obstructive sleep apnea. (Was he amenable to physical examination?) Because he had relatively few medical problems, he was deemed suitable for day surgery with the possibility of hospital admission if he developed perioperative complications.

An anesthetic plan was formed with the assistance of his parents. Prior to the start of the case, a team huddle consisting of the anesthesia consultant, resident, anesthetic nurse, scrub nurse and attendant was held to brief everyone on the action plan. Two extra male attendants were enlisted to help with restraining the patient as necessary. A special exception was made to allow both parents to be present during anesthetic induction. Both intravenous (IV) and intramuscular (IM) doses of succinylcholine were calculated and prepared in case the patient went into laryngospasm before the IV access was secured.

Our patient refused topical local anesthetic cream on his hand. Oral premedication with 10 mg midazolam and 100 mg ketamine was administered; even though he spat out some, there was some sedative effect. Twenty minutes later, he was passive enough to be moved into operating theatre on a wheelchair accompanied by both parents. Inhalational induction was started with 100% oxygen rather than 50% nitrous oxide and oxygen mix to increase lung stores of oxygen in anticipation of

a longer time needed for airway manipulation. Gradual increments of sevoflurane were administered till 8% via an Ayre's T-piece. IV access was secured in one attempt. He was then lifted from the wheelchair to the operating table by five people. Face mask ventilation was confirmed to be possible before administering IV atracurium 0.5 mg/kg. Co-phenylcaine spray was applied to both nostrils to reduce the risk bleeding during nasal intubation. After 3 min, direct laryngoscopy was attempted which showed a grade 1 larynx. Oral intubation with a size 6 cuffed endotracheal tube (ETT) was done to confirm the appropriate size and allow for further pre-oxygenation before the subsequent successful nasal intubation with the same sized ETT.

IV paracetamol 15 mg/kg, IV fentanyl 2 µg/kg and local anesthetic were given for multimodal analgesia. IV dexamethasone 4 mg and IV ondansetron 4 mg were given for antiemesis. Surgery concluded uneventfully 120 min later. Neuromuscular block was reversed with neostigmine 0.05 mg/kg and atropine 0.02 mg/kg without any arrhythmias noted. In the post anesthetic care unit (PACU), he was reunited early with his parents. He was discharged home after 6 h of uneventful observation.

Discussion

In intellectually challenged children with behavioral issues, hospital admission can be stressful for the family and child. Therefore, day surgery should be considered where possible. Whether a procedure can be done as day surgery or not should involve an evaluation by an anesthetist. The successful management of this patient as a day surgery patient depended on multidisciplinary collaboration. It is important to emphasize that not every patient with RTS should be considered for day surgery and regardless; the procedure should be performed in a center with admission facilities for post anesthetic or surgical complications that may arise. For this reason, this patient was scheduled to have his procedure done in a tertiary pediatric hospital rather than a stand-alone outpatient facility.

A normal airway assessment does not preclude the possibility of a difficult airway. There are some reports of difficult mask ventilation and laryngoscopy in RTS, primarily due to high arched palate, hypoplastic mandible and limited mouth opening [6, 7]. Specific to the nasal region, choanal atresia or nasal septum deviation can make nasal intubation more difficult. Nasal bleeding can develop and complicate reintubation. The presence of a vascular ring, congenital tracheal stenosis, laryngomalacia may predispose to airway obstruction. Therefore, if there is any clinical suspicion, fiberoptic endoscopy may be considered to evaluate these passages.

In cases where the risk of difficult intubation outweighs the risk of aspiration, we suggest ensuring mask ventilation is possible before paralyzing the patient. Rapid sequence intubation, if needed, should ideally be done with rocuronium with sugammadex on standby. There should also be thought given to the backup plan should laryngoscopy fail. Supraglottic devices like laryngeal mask airways have been used successfully in these cases [5].

Since our patient had no gastroesophageal reflux issues

and would not cooperate with IV plug setting, we chose inhalational induction and maintained spontaneous ventilation until IV access was obtained. Recognizing that nasal intubation is usually more difficult than oral particularly in children, sizing of the ETT may be difficult for children who are small for age and “in-between sizes”, and that patients with RTS are at higher risk of desaturation from various causes, we intubated our patient orally first attempting nasal intubation. This avoided unnecessary nasal trauma and intubation attempts due to a wrongly sized ETT and ensured that our patient did not desaturate throughout intubation.

For children with special needs such as intellectual disability or behavioral issues, every hospital visit is potentially stressful for the child particularly if repeated hospitalization visits or admissions are required because of associated medical conditions. If an anesthetic encounter is planned well with the caregivers on board, taking into consideration the child’s likes or dislikes, sensory intolerance and anxieties, then by working around them, a more pleasant experience may reduce the anxiety during subsequent encounters. Communication, parental presence and premedication served important roles in our child’s inhalational induction. Forceful physical restraining techniques should be avoided if possible as these could have negative psychological impact and increase subsequent perioperative anxiety. The patient’s father was considerably anxious before the anesthetic induction but expressed his appreciation afterwards that this experience was not as emotionally and psychologically traumatic compared to his child’s previous medical encounter.

In situation where potential crises could occur, a team brief is useful to delineate roles, go through contingency plans and improve patient safety [15]. Communication failures account for 43% of errors in the operating theatre in the USA [16]. The team brief promotes a shared mental model of what is expected to happen so that the cognitive resources of the team are leveraged for early error detection when deviations from the plan occur. Team briefing should not only be limited to the medical personnel: we suggest that a briefing be conducted for parents as well. Parental presence during induction of anesthesia has been shown to have controversial results in anxiety and children cooperation improvement [17]. Calm parents may improve preoperative anxiety, while highly anxious parents do not. The objectives of preoperative parental briefing, especially for complex patients should include: 1) Familiarization with the child’s signals of anxiety and coping behavior (violent reactions should be anticipated); 2) Deciding the extent of parents’ participation at induction and recovery; and 3) Educating parents on what to expect as their child recovers from anesthesia including the management of emergence delirium. This in turn allows the planning and implementation of an anesthetic plan tailored to a child’s needs, reduces the parents’ anxiety at their child’s behavior during recovery from anesthesia, and increases patient and parental overall satisfaction with the experience.

Conclusions

In summary, RTS is a rare genetic disorder that has implica-

tions for anesthesia. Such patients may be at increased risk of anesthetic complications. These patients need to be evaluated by an anesthetist on their suitability for day surgery and the procedure should be done in a facility capable of managing difficult airways with inpatient services. Perioperative planning and close multidisciplinary communication are essential to the successful management of these patients, particularly if they require repeated hospital visits. The parents should be engaged and briefed on how they can work with healthcare workers to make it less stressful for their child during repeated hospital visits.

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Financial Disclosure

None to declare.

Conflict of Interest

None to declare.

Informed Consent

Informed consent was obtained from the patient’s father.

Author Contributions

PSAY drafted and reviewed manuscript; HLEL reviewed manuscript.

Data Availability

Any inquiries regarding supporting data availability of this study should be directed to the corresponding author.

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