

ANESTHETIC MANAGEMENT OF CHILDREN WITH RUBINSTEIN-TAYBI SYNDROME

- Case Reports -

SHVETANK AGARWAL, MD, YASSER HAIDAR AHMAD, MD,
MONICA TALPESH, MD AND MARIA ZESTOS, MD

Abstract

A limited number of cases of anesthetic management of Rubinstein-Taybi Syndrome (RTS) have been reported since this syndrome was first diagnosed in 1963. After some well-publicized complications following anesthesia for patients with RTS, there has been great interest in avoiding all precipitant factors and careful monitoring during intraoperative and postoperative periods.

This case series examines the cases of three pediatric patients with RTS who presented to the Children's Hospital of Michigan for different surgeries. We aim in this study to share information about this rare syndrome and to emphasize how this case series allowed us to improve our anesthetic management. In each case, we adjusted our techniques using information from preceding cases to avoid complications in the following encounter.

Introduction

RTS is a rare congenital anomaly that affects many organ systems of the body with a frequency of approximately 1 in 100,000 newborns¹. The findings associated with this syndrome include growth retardation, delayed bone age, mental retardation, abnormally broad thumbs and great toes, breathing and swallowing difficulties. Most affected children experience delays in attaining developmental milestones and delays in the acquisition of skills requiring coordination of muscular and mental activity (psychomotor retardation). In addition, many individuals with RTS may have malformations of the heart, kidneys, urogenital system, and/or skeletal system. The abnormalities of the head and face include widely spaced eyes (hypertelorism), a broad nasal bridge, an abnormally large or "beak-shaped" nose, and an unusually small, hypoplastic lower jaw (micrognathia) with small mouth opening. The classical facial appearance is well-established, striking and easy to recognize.

Due to their craniofacial abnormalities, delayed development and gastroesophageal reflux disease (GERD), these patients are prone to difficult intubation and airway compromise². These features may complicate not just the intraoperative anesthetic management, but also cause problems in the immediate and late postoperative period.

Department of Anesthesiology, Children's Hospital of Michigan/Wayne State University, 3901 Beaubien Blvd, Detroit, MI 48201, USA.

Corresponding author: Shvetank Agarwal, MD. Tel: 313-745-7233, Fax: 313-993-3889, E-mail address: sagarwal@med.wayne.edu

Case 1 A

The first patient was diagnosed with RTS at 2 weeks of age, after unremarkable term delivery. He had typical broad thumbs, broad first toes, large forehead, low set ears, anti-mongoloid slant of the palpebral fissures, bilateral undescended testes, bilateral renal calculi, renal tubular acidosis, cholelithiasis, hypotonia, mild GERD, and severe developmental delay. His echocardiogram showed a small patent foramen ovale, mild mitral valve regurgitation with preserved ventricular function. There were no features suggestive of abnormal airway.

At 9 months of age and a weight of 4.5 kg, he presented for MRI of the spine under general anesthesia. No difficulty in intubation was encountered with a Miller 0 laryngoscope blade and a 4.0 uncuffed endotracheal tube (ETT) and the patient had an uneventful intra-procedural course. There was no mention in the anesthesia record of any audible leak heard around the ETT. The anesthesia consisted of isoflurane, 1 mcg/kg of fentanyl and 0.5 mg of pancuronium. After a short stay in the postoperative care unit, he was discharged home. The following day, he developed respiratory distress necessitating a visit to the emergency department where he was found to have copious secretions. Chest x-ray showed no evidence of pneumonia. After thorough suctioning of the airway and several hours of observation, the patient improved and was sent home.

Case 1 B

The above patient presented a month later at 10 months of age and a weight of 4.8 kg for laminectomy and release of tethered cord. The patient was induced with 50% N₂O/O₂ and 8% sevoflurane using a face mask. An IV access was started and propofol 2 mg/kg, fentanyl 2 mcg/kg and vecuronium 0.1 mg/kg were given. On this occasion a smaller sized tube was chosen to minimize the likelihood of airway trauma, edema, or tracheomalacia. Gentle mask ventilation was followed by an easy intubation with a 3.5 uncuffed ETT using a Miller 0 laryngoscope blade and secured at 11 cm after confirmation of bilateral air entry. No audible leak was noted with the smaller tube size, which suggested that the size 4 ETT used in the previous anesthetic in this patient one month was inappropriately large. The patient was turned prone and anesthesia was maintained with isoflurane in 50% O₂/air and propofol infusion at 150 mcg/kg/min. No further doses of neuromuscular blockers were given.

The surgery lasted approximately 4.5 hours. The trachea was extubated after the patient was fully awake and after thorough suctioning of his airway. His postoperative course was uneventful and was discharged home on postoperative day 3.

Case 2

A 10 year old boy presented for dental restoration because of advanced dental caries. Our patient showed many of the typical craniofacial features of RTS and significant developmental delay. Examination showed a boy of short stature weighing 30 kg. He had a prominent broad nasal bridge, arched thick eyebrows and a prominent beaked nose. No obvious anomaly of the maxilla or upper jaw was noted. However, the patient had a small mouth and adequate but limited mouth opening which may reflect poor development and malpositioning of the mandible or lower jaw that is often seen with RTS.

Before induction, we made sure that equipment necessary to manage difficult airway were immediately available. We induced anesthesia with 70% N₂O/O₂ mixture in sevoflurane 8% via face mask and intubated using a smaller tube than normal based on our experience in the previous case. His heart rate was 145 bpm, SpO₂ 97% and BP 110/60 mmHg. After IV access was obtained 1 mcg/kg of fentanyl and 0.05 mg/kg of vecuronium were injected.

Because choanal atresia and a deviated nasal septum has been described in RTS, the nasal mucosa was well prepared with oxymetazoline (Afrin). A red rubber suctioning tube was used as a guide for the 5.5mm nasal RAE tube to assess the patency of the naris and to avoid trauma. The right naris was found to be obstructed however; the tube was easily advanced in the left naris. At laryngoscopy, a very high arched palate was confirmed and the cords were partially seen (Cormack Grade II). Intubation was uneventful.

Anesthesia was maintained with isoflurane 1.5% in 50% oxygen/air. Interestingly, there were three documented episodes of self-limiting oxygen desaturations to 90% without any provocation. The surgery lasted 2½ hours. To avoid the risk of arrhythmia, anticholinesterase was not given, with no evidence of residual muscle relaxant effect. Good airway and gastric suctioning was performed and the patient was extubated ten minutes later when fully awake without any complications.

Case 3

A 4 year old boy presented for dental restoration because of advanced dental caries. He had many craniofacial features of RTS and global developmental delay. Again, equipment necessary for difficult airway were immediately available. After uneventful mask induction with 70% N₂O/O₂ mixture and sevoflurane, the nasal mucosa was prepared with oxymetazoline (Afrin). Despite this preparation there was bleeding in the posterior pharynx that obscured the view of the vocal cords and led to failed intubation on first attempt. After repeated suctioning, intubation was successful at the third attempt. Anesthesia was maintained with isoflurane 1.5% in 50% oxygen/air. Opioids were kept minimal with a total dose of 2 mcg/kg fentanyl IV. Good airway and gastric suctioning was performed and the patient was extubated when fully awake without any complications.

Discussion

In this case series we collected information about RTS and our anesthesia techniques for four anesthetics and the following are the lessons learned:

1. Copious secretions causing airway compromise has been reported in literature and was seen in our case#1A³. We resorted to careful oropharyngeal and endotracheal suctioning prior extubation to avoid this complication in the subsequent three cases. We also confirmed accurate placement to avoid endobronchial intubation during changes in patient positioning and subsequent atelectasis.
2. Intraoperative self-limiting desaturations to 90% were seen in case #2 without any provocation and these have been reported by Critchley et al⁴. Respiratory infections and complications are major causes of morbidity and mortality in the first years of life⁵. Studies have shown that breathing disturbances occur in 11% of subjects with RTS⁶.
3. Airway narrowing and tracheomalacia is always a possibility and could have been the cause of

postoperative airway compromise in case 1A. Hannekam et al have reported abnormal upper and lower airway narrowing in non-anesthetized subjects with RTS⁶. To avoid airway trauma and tracheomalacia, we deliberately used a relatively smaller sized endotracheal tube in our patients.

4. Cardiovascular abnormalities are not uncommon in these patients⁵. In case #1B, we carefully avoided bradycardia and increased afterload in order to prevent worsening of mitral regurgitation fraction and subsequent reduction of the cardiac output. Our patient also had a patent foramen ovale, so we were very vigilant in avoiding air bubbles in the intravenous line to prevent paradoxical emboli. These cardiac abnormalities also predispose to arrhythmias with the use of arrhythmogenic drugs like succinylcholine, atropine and neostigmine³. We therefore avoided the use of the aforementioned drugs and our patients did not have any arrhythmia at any time during the hospital stay.
5. Epsitaxis complicating airway management occurred in case #3. Though this cannot be labeled as difficult intubation, yet in these patients a difficult airway should always be anticipated and an emergency airway cart including a fiberoptic bronchoscope should be available, despite history of easy intubations during prior anesthetics in the same patient.
6. Delayed recovery after general anesthesia in these patients is also a concern and has been reported in literature⁷. In our case series, we did not encounter this problem, though we were well prepared for it.

Conclusion

Children with RTS vary widely in the range and severity of symptoms and physical findings as well as the complications developed during the perioperative period. Whatever the phenotypic presentation of patients with RTS, the physical challenges to the anesthesiologist should always be centered about three main areas: craniofacial anomalies, airway and pulmonary complications and cardiac anomalies. We strongly suggest careful planning of the anesthetic and airway management with close observation and monitoring extending well into the postoperative period.

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