Turk J Anaesthesiol Reanim 2016; 44: 152-4 DOI: 10.5152/TJAR.2016.76992

Case Report

Anaesthetic Management of Children with Rubinstein-Taybi Syndrome

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Abstract

Rubinstein-Taybi syndrome (RTS) is a rare, autosomal dominant syndrome presenting with mental retardation and physical abnormalities, including broad thumbs, big and broad toes, short stature and craniofacial anomalies. Special attention was paid to the possibilities of difficult airway, aspiration pneumonia and cardiovascular dysfunction during anaesthesia. Micrognathia, retrognathia, broad nasal bridge, abnormally large or 'beak-shaped' nose, hypoplastic maxilla and small mouth-typical dysmorphic facial features are one of the biggest causes of the difficult airway in this syndrome. Approximately one-third of the affected individuals have a variety of congenital heart diseases. Recurrent respiratory infections are likely to be the result of microaspiration or gastro-oesophageal reflux in this syndrome. In this case report, we discussed the anaesthesia management of a child with RTS who underwent right endoscopic dacryocystorhinostomy.

Keywords: Rubinstein-Taybi syndrome, airway management, general anaesthesia

Introduction

Rubinstein–Taybi syndrome (RTS) is a rare autosomal dominant-inherited syndrome in which a characteristic facial appearance, broad thumbs and first toes are observed (1). The incidence of the syndrome, first defined in 1963 by Jack Herbert Rubinstein and Hosshang Taybi, is 1/250,000–300,000, and it is equally observed in both sexes. At present, two genes have been identified to be responsible for the disease: abnormalities related to the CREBBP gene were detected in more than 60% of patients and those related to the EP300 gene were detected in 3% of patients (2).

Growth retardation, delayed bone age, mental retardation and breathing and swallowing difficulties are among the clinical symptoms associated with the syndrome. In addition, multiple genetic defects such as short stature, microcephaly, micrognathia, hearing loss, skin problems, congenital heart abnormalities, cryptorchidism and renal anomalies are among the common symptoms. Major causes of deaths with the syndrome, particularly during the first year, are aspiration pneumonia and hereditary heart disease (3).

Anaesthetic management in patients with RTS has several difficulties. The most important challenge for anaesthesia applications in RTS is difficult intubation, which is observed depending on craniofacial defects. Besides, respiratory infections and heart rhythm disorders make the anaesthesia applications difficult in these patients (4). In our case report, accompanied with the literature, we aimed to present the anaesthesia application in an RTS-diagnosed patient for whom right endoscopic dacryocystorhinostomy was planned.

Case Presentation

Right endoscopic dacryocystorhinostomy was planned under elective conditions for a 14-year old male who weighed 45 kg, was 140 cm tall and had first-degree mental retardation with the diagnosis of RTS. In the physical examination, the presence of microstomia, macroglossia, narrow palate, micrognathia and retrognathia was detected. The Mallampati score was found to be 3 in the airway assessment performed for intubation. The patient did not have any limitations in the flexion or extension of the neck. Results of biochemical tests and a complete blood count in the laboratory tests were within normal limits. After the evaluation of paediatric cardiology, no pathology was found in the heart. No additional suggestions were made in result of the consultation with paediatric neurology. Considering the patient's Mallampati, airways, endotracheal tubes, stylettes, Miller and Macintosh blades and LMAs of various sizes and appropriate for his age and weight were prepared for

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Received : 15.10.2015
Accepted : 14.12.2015

difficult intubation. The Department of Otolaryngology was consulted, and preparation was made for the tracheostomy if necessary. Written informed consent required for the anaesthesia was taken from his parents.

Intravenously (iv), 2 mg of midazolam and 10 mg of ranitidine were administered to the patient who was taken to the operating room for premedication purposes. After ECG, oxygen saturation and non-invasive blood pressure monitoring was performed in the patient; 2 mg kg^{-1} propofol and 1 μg kg^{-1} remifentanil were intravenously used for anaesthesia induction. In addition, 0.6 mg kg^{-1} rocuronium was used as a muscle relaxant in the patient, who had no problems with the ventilation with the mask. The insertion of the No. 5.5 spiral tube was planned in the patient for endotracheal intubation, and intubation was achieved with a video laryngoscope in the second trial. Minute ventilation was set according to the end-tidal carbon dioxide ($ETCO_2$) value. Anaesthesia was maintained with 2% sevoflurane, 50% oxygen and 50% dry air. The operation lasted for about 90 min, and the perioperative haemodynamic parameters were maintained as stable. After the surgery, the patient, in whom 2 mg kg^{-1} sugammadex was intravenously administered and whose muscle strength returned to a sufficient amount, was extubated. The patient, who had adequate spontaneous respiration and normal haemodynamic parameters, was then taken to the postanaesthesia care unit. The patient's vital signs maintained as stable; therefore, he was transferred to the service after full recovery was ensured. The patient did not experience any complications and was discharged on the second postoperative day.

Discussion

Rubinstein–Taybi syndrome is a hereditary disease with an incidence of as rare as 1/250,000–300,000, but it can be as common as 1/600 in patients who are followed due to mental retardation (5). The majority of the surgeries that RTS patients undergo are orthopaedic, orthodontic, ophthalmic or heart surgery. In terms of anaesthetic management of the syndrome, the most important features are the airway management challenges, the risk of aspiration pneumonia and cardiovascular pathologies (6).

Patients with RTS are considered as a potential patient group with difficult intubation. Micrognatia, retrognathia, a broad nasal bridge, an abnormally large or 'beak-shaped' nose, a narrow high-arched palate, a hypoplastic maxilla and a small mouth opening are considered as the main craniofacial reasons for laryngoscopy; these factors cause intubation difficulty in this patient group (3). In the literature, alternative methods such as nasal intubation (5), intubation accompanied by fibreoptic bronchoscopy (7), ProSeal LMA use (8) and an air-Q intubation laryngeal mask (air-Q ILA) (9) have been used in addition to oral endotracheal intubation in the airway approaches and management of these patients. However, before using these methods, the presence of a vascular ring that occurs in the nasal region due to choanal atresia, nasal septum deviation, lower airway narrowing, postcricoid web, laryngomalacia and tracheoesophageal compressions should carefully be questioned in a detailed airway examination (10). After the operations performed in the nasal region, sufficient nasal bleeding may develop to prevent intubation (3). Therefore, nasal anatomy and the passage should be assessed prior to the nasal intubation and fibre-optic intubation.

Another important feature of the syndrome that may affect the airway management is the risk of aspiration. Hereditary tracheal stenosis, abnormal pulmonary lobulation, excessive mucus secretion and gastroesophageal reflux increase the risk of aspiration in these patients (11). Gastroesophageal reflux that begins in the first year of life may reduce the patient's nutrition and cause developmental retardation. Recurrent infections resulting from microaspiration in the respiratory tract and gastroesophageal reflux may be seen in about 75% of RTS patients. LMA use for the purpose of airway management is controversial in such patients. When the cases in which LMA was used were analysed in the literature, although the patients stated that they did not have any existing gastroesophageal reflux problems, the usage of ProSeal LMA was preferred instead of classic LMA in these cases (8, 12). In such cases, ProSeal LMA enables aspirating the stomach content through the gastrointestinal tract using an orogastric tube as well as reducing the likelihood of failed or insufficient ventilation (13). Air-Q ILA, which is easy to implant and implement and provides a good channel for the intubation, may be preferred in patients with reflux and in whom intubation could be difficult (9).

Cervical hyperkyphosis can be seen in 62% of patients with RTS, while scoliosis can be seen in 38%. Such vertebral anomalies may make it difficult to provide a suitable position during intubation and airway control in patients (14).

In the case we presented, the presence of macroglossia, a narrow palate, micrognathia and retrognathia, which are the typical craniofacial features of the syndrome, was detected. In airway evaluations made for intubation, it was observed that the Mallampati score was class 3. Intubation was successfully performed with the help of video laryngoscope in the patient, who did not have aspiration pneumonia in the anamnesis.

Heart defects are present in 24–38% of patients with RTS. The large part of anomalies first identified in patients with RTS in 1964 consisted of isolated lesions, such as the atrium septum defect, ventricular septum defect, patent ductus arteriosus, coarctation of the aorta, pulmonary valve stenosis, bicuspid aorta, aortic valve stenosis, vascular rings and conduction abnormalities. In a rare part, complex hereditary heart lesions, such as hypoplastic left heart syndrome, were described (15).

In patients with heart problems, the use of anticholinesterase and anticholinergic drugs may increase the risk of arrhythmia. The sympathetic and parasympathetic system changes formed by, in particular, neostigmine and atropine, which are

the drugs used in anaesthesia, can trigger ectopic rhythms. Because it causes supraventricular tachycardia with premature atrial and ventricular contractions, succinylcholine should be avoided in this patient group (4-6). No pathology was found in our patient in the results of the preoperative paediatric cardiology consultation. Rocuronium was used as a non-depolarizing muscle relaxant in the induction of anaesthesia. Before the extubation of the patient, sugammadex was preferred rather than neostigmine and atropine. No preoperative and postoperative problems were encountered.

Conclusion

The control of a difficult airway and difficult intubation in terms of anaesthesia management is the most important problem in patients with RTS because of their abnormal anatomical structures. Therefore, it is necessary to prepare for difficult ventilation and intubation prior to anaesthesia. Because succinylcholine, neostigmine and atropine will increase the risk of arrhythmia, particularly in patients with heart defects, the use of a rocuronium–sugammadex combination may be a good alternative.

Informed Consent: Written informed consent was obtained from patient's family who participated in this case.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – M.A.K.; Design – M.A.K., H.S.; Supervision – Z.A, B.A.; Resources – M.A.K., B.A.; Materials – Z.A., M.A.K.; Data Collection and/or Processing – M.A.K., Z.A., H.S.; Analysis and/or Interpretation – M.A.K., H.S., Z.A., B.A.; Literature Search – M.A.K., B.A.; Writing Manuscript – M.A.K., B.A.; Critical Review – Z.A., B.A., H.S.; Other – M.A.K., Z.A.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

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