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CLINICAL REPORT

Anesthetic management of pediatric patients with Emanuel syndrome

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Abstract Emanuel syndrome is a rare anomaly associated with multiple systemic malformations. We present two cases involving pediatric patients with Emanuel syndrome. The first patient presented with micrognathia and had patent ductus arteriosus and a single kidney. The patient was difficult to intubate with McGRATH® but was successfully intubated with an Airtraq® device. The second patient did not present with micrognathia and was not difficult to intubate. A thorough examination of the heart, kidney, and spinal cord is important when planning the anesthetic management of patients with Emanuel syndrome. Moreover, adequate preparation for a difficult airway is essential, and the Airtraq® device may be useful for intubating patients with Emanuel syndrome with micrognathia.

Keywords Emanuel syndrome · Anesthesia · Micrognathia · Difficult airway · Airway management · Airtraq · Supernumerary derivative 22 syndrome

Introduction

Emanuel syndrome (ES) is a rare anomaly characterized by facial dysmorphism, micrognathia, congenital heart disease, renal anomalies, and mental retardation. It is caused by the presence of a supernumerary derivative chromosome consisting of portions of chromosome 11 and chromosome 22. The anesthetic management of patients with ES is challenging because of micrognathia which can cause upper airway obstruction and unsuccessful intubation, and congenital heart disease which can cause hemodynamic instability. Other anomalies such as renal malformations or spinal cord anomalies potentially affect the anesthetic management. Here, we report two cases involving children with ES and discuss important clinical considerations. Written consent to publish this case series was obtained from both parents of each child. The clinical features of ES have been reported in many articles; however, to the best of our knowledge, there is only one case reporting the anesthetic management of ES [1].

Case 1

This male pediatric patient underwent surgery under general anesthesia four times. He was delivered normally at 40 weeks gestation (birth weight 2,770 g). A clinical examination revealed micrognathia, malformed ears, ear pits, stenosis of the proctodeum, membranous anal atresia, and a sacral dimple. A further work-up revealed a cleft palate, single kidney, and an atrial septal defect (ASD). No spinal cord abnormality was detected on ultrasound examination. At the age of 13 days and weighing 2.7 kg, he underwent a surgical ligation for patent ductus arteriosus under general anesthesia due to exacerbating symptoms of heart failure. Anesthesia was induced intravenously with midazolam and fentanyl, and there was no difficulty in mask ventilation or tracheal intubation. Soon after extubation on the fourth postoperative day, he developed upper airway obstruction caused by glossoptosis and required oxygen administration. His renal function deteriorated after surgery and was considered to be associated with the presence of a single kidney and the use of diuretics. The renal function recovered after diuretic administration was discontinued. He was diagnosed with ES at the age of 1 month based on a chromosome study.

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At the age of 1 year and 6 months and weighing 11.7 kg, tympanostomy tube insertion was performed under general anesthesia induced via a mask using sevoflurane. The face mask ventilation was uncomplicated; however, the tracheal intubation by direct laryngoscopy failed because of poor laryngeal visualization (Cormack-Lehane grade 4). We then tried to intubate with an Airtraq® laryngoscope; this enabled complete glottis visualization, and tracheal intubation was successful. After extubation, a nasopharyngeal airway was inserted for a short period because of severe upper airway obstruction.

At the age of 2 years and 1 month and weighing 11.8 kg, the patient underwent laparoscopic gastrostomy. Direct laryngoscopy with a Macintosh size 2 blade failed to visualize the epiglottis (Cormack-Lehane grade 4). A McGRATH® MAC video laryngoscope with a size 2 blade was then used for intubation, but failed to visualize the glottis. Finally, tracheal intubation was successfully performed using a size 2 Airtraq® device.

The patient underwent cleft palate repair at the age of 2 years and 3 months and weighing 11.9 kg. Although his laryngeal view score had improved to grade 3, tracheal intubation by direct laryngoscopy failed. Again, tracheal intubation was successful with a size 2 Airtraq® device. After palatoplasty, a nasopharyngeal airway was placed by the plastic surgeon to prevent upper airway obstruction for 3 days, as is the routine practice at our hospital.

Case 2

A female pediatric patient underwent surgery under general anesthesia twice—for palatoplasty at the age of 1 year and 2 months and weighing 8.7 kg, and for cheiloplasty at the age of 1 year and 5 months and weighing 8.9 kg. She was born at term (birth weight 2,383 g) with facial dysmorphism including bilateral microtia, atresia of the right external auditory meatus, cleft palate, and left macrostomia. She was also found to have an ASD with mild pulmonary artery stenosis. Although Goldenhar syndrome was suspected based on the clinical presentation, she was diagnosed with ES based on a chromosome study at the age of 4 months. She snored because of laryngomalacia and glossoptosis during her infancy; however, her symptoms improved gradually.

General anesthesia was induced both times via a mask using sevoflurane in oxygen and nitrous oxide. There was no difficulty in mask ventilation or tracheal intubation in either instance. Direct laryngoscopy with a Macintosh size 2 blade provided a full view of the glottis. She was extubated and recovered uneventfully. As the ASD was small and her pulmonary artery stenosis was mild, specific management was not necessary during surgery.

Discussion

ES is characterized by multiple congenital anomalies and complications caused by the presence of a supernumerary derivative chromosome. We briefly summarize below the anesthetic management of ES on the basis of bibliographic consideration.

Micrognathia/Pierre Robin sequence

As micrognathia and Pierre Robin sequence have been reported in 60 and 34 % of patients with ES, respectively [2], it is essential to prepare for difficult airway management in ES patients. In Case 1, direct laryngoscopy was difficult and tracheal intubation was unsuccessful, except during the patient's first operation. We successfully intubated his trachea using an Airtraq® three times. In the pediatric anesthesia literature, there are some reports on the successful use of an Airtraq® in a child with Treacher Collins syndrome [3, 4] and a child with Pierre Robin syndrome [5, 6]. Therefore, we believe that the Airtraq® is useful for management of a difficult airway in children with ES. During his third operation, we tried to intubate his trachea using a McGRATH® device, but we were unable to visualize the vocal cords because the shape and length of the blade did not fit the configuration and size of his mandible. To date, there is no report describing the use of a McGRATH® device in children with a difficult airway. Moreover, a recent manikin study [7] revealed that the success rate of intubation with the Airtraq® was higher than with a McGRATH® in a difficult airway model. The Airtraq® does not require a large mandibular space to align the direction of its line of vision with the laryngeal axis because of its steeply curved blade (an approximately 90° angle). On the other hand, the McGRATH® requires a certain volume of mandibular space to align the direction of its line of vision with the laryngeal axis because of the less extreme angle of the blade. We believe that the Airtraq® is superior to the McGRATH® for the intubation of pediatric patients with severe mandibular hypoplasia.

According to the ASA guideline, video-assisted laryngoscopy is recommended prior to intubation for determining an anticipated difficult airway, especially in adults [8]. Although this guideline does not include a recommendation for difficult airway management in children, we consider the Airtraq® could be recommended as an initial approach to intubation for pediatric patients with micrognathia.

In a previous report of the anesthetic management of ES [1], tracheal intubation failed using direct laryngoscopy, but was successful using a fiberoptic-guided technique through a laryngeal mask airway. However, the video-assisted laryngoscopy was not used in the previous report, because

it was not available for children in 2003. We have added the new information about the usefulness of video-assisted laryngoscopy for ES.

Congenital heart malformation

Congenital heart malformation is frequently associated with ES. The most common malformations are ASD, ventricular septal defect, and patent ductus arteriosus. Other malformations include coarctation of the aorta, pulmonic stenosis, and total anomalous pulmonary venous return [2, 9]. Although Case 1 had ASD and Case 2 had both ASD and pulmonary artery stenosis, additional monitoring such as arterial or central venous pressure monitoring was not required. A thorough examination should be performed before surgery, and anesthesiologists should pay attention to the hemodynamic status of each heart defect.

Renal malformations

Renal malformations were reported to occur in 19–36 % of patients with ES [2, 10]. Recently, reported cases of renal malformations have increased because of the improvement in imaging techniques. Perioperative monitoring of renal function is needed for ES patients because they have a potential risk of renal dysfunction, similar to the one that occurred in Case 1, where stringent monitoring enabled us to detect and treat the renal dysfunction early.

Sacral dimple

A sacral dimple is reported in 24 % of patients with ES [2]. A dimple may represent underlying malformations of the spine, especially spina bifida occulta [11]. As there is a risk of neural injury in performing neuraxial block for patients who have cutaneous abnormalities of the back, it is necessary to confirm the presence or absence of a sacral dimple for patients scheduled for spinal or epidural blocks. It is also necessary to perform an ultrasound examination or magnetic resonance imaging for patients with a sacral dimple. In Case 1, the patient who had a sacral dimple underwent an ultrasound examination which showed a spine with a normal appearance.

We experienced 2 cases of ES with multiple anomalies undergoing general anesthesia. A thorough examination of the heart, kidney, and spinal cord is important before general anesthesia, and adequate preparations should be made for airway treatment because of the high risk of difficult airway and upper airway obstruction. The Airtraq® device may be more useful than the McGRATH® device to intubate ES patients with micrognathia.

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