Anesthetic Management of a Patient With Emanuel Syndrome

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Emanuel syndrome is associated with supernumerary chromosome, which consists of the extra genetic material from chromosome 11 and 22. The frequency of this syndrome has been reported as 1 in 110,000. It is a rare anomaly associated with multiple systemic malformations such as micrognathia and congenital heart disease. In addition, patients with Emanuel syndrome may have seizure disorders. We experienced anesthetic management of a patient with Emanuel syndrome who underwent palatoplasty. This patient had received tracheotomy due to micrognathia. In addition, he had atrial septal defect, mild pulmonary artery stenosis, and cleft palate. Palatoplasty was performed without any complication during anesthesia. Close attention was directed to cardiac function, seizure, and airway management.

Key Words: Emanuel syndrome; General anesthesia; Congenital heart disease.

Emanuel syndrome is a rare recessive hereditary disorder, first described in 1976.[1] Most of the clinical information was published in the 1980s.[2] The features include tracheomalacia, micrognathia, seizure disorder, and congenital heart disease.[3,4] However, anesthetic management of these patients has been only minimally reported.[4,5] We report a case of a patient with Emanuel syndrome undergoing palatoplasty under general anesthesia.

CASE REPORT

The patient was a 2-year-old boy, 82.3 cm (32.4 in) in height and 9.8 kg in weight. He was diagnosed with Emanuel syndrome on the basis of chromosomal study when he was an infant. His clinical features were tracheomalacia, micrognathia, atrial septal defect (ASD), mild pulmonary valve stenosis (PS), cleft palate, preauricular tags, severe hearing loss, and microtia. He had received tracheotomy at 9 days old due to micrognathia with breathing difficulty. At the time of palatoplasty surgery, his tracheostomy was still in place. He could not walk by himself, but could turn himself over. He spoke only a few words and was uncooperative for preanesthetic evaluation. Palatoplasty under general anesthesia was scheduled for primary closure of the cleft palate.

Preoperative echocardiography revealed a small ASD (12.7×4.7 mm) with left to right shunt and PS (pressure gradient, 24 mm Hg). Ejection fraction was 67%. A 12-lead electrocardiogram (ECG) revealed sinus rhythm. Electrolytes were within normal limits. No abnormal findings were observed on chest X-ray.

On admission, his heart rate (HR) was 123 bpm, blood pressure (BP) was 105/70 mm Hg, and oxygen saturation by pulse oximetry (SpO_2) was 100% on room air. On the day of surgery, no premedication was given, and he was transferred to the operating room. Anesthesia was induced with inhalation of sevoflurane 5–8% in oxygen after the start of noninvasive

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monitoring for SpO_2 (100%). After the loss of consciousness, mask ventilation was easy. Monitoring of ECG (sinus rhythm), BP (105/75 mm Hg), and HR (120 bpm) was started in addition to Bispectral Index (BIS) monitoring. In addition, stroke volume (SV): 9.6 mL, stroke volume variation (SVV): 16%, and cardiac output (CO): 1.2 L/min were also monitored by using Aesculon®. Fentanyl 10 mcg, atropine 50 mcg, and rocuronium 6 mg were administered after peripheral intravenous access was obtained. The tracheostomy tube was exchanged for a coiled tube after induction. Anesthesia was maintained with sevoflurane 1–3% in air and oxygen. In addition, a total of 80 mcg of fentanyl was administered intermittently for analgesia. Approximately 15 minutes after tube was exchanged, cardiac functions drastically changed (BP: 95/62 mm Hg, HR: 150 bpm, SV: 15.5 mL, SVV: 14%, CO: 2.2 L/min), but it gradually recovered to the baseline level in 30 minutes. From that point, stable hemodynamics and ventilation were maintained during surgery. BP was maintained at 90–110/40–65 mm Hg, HR was 122–130 bpm, and end-tidal carbon dioxide ($EtCO_2$) was 35–40 mm Hg. Stable cardiac parameters were also maintained (SV: 9.7–12.2 mL, SVV: 9–11%, CO: 1.1–1.2 L/min). The palatoplasty was completed in 7 hours, 43 minutes without any surgical and/or other anesthetic problems. There was minimal blood loss during operation and he received a total of 400 mL lactated Ringer's solution with 1% glucose. Urine volume was 45 mL. He emerged from general anesthesia 10 minutes after discontinuing inhalation of sevoflurane and was extubated awake. There were stable hemodynamics (BP: 105/58 mm Hg, HR: 135 bpm, SpO_2: 98%), and no adverse neurological symptoms were observed.

DISCUSSION

Emanuel syndrome is associated with excess chromosomal material, which consists of redundant genetic material from chromosomes 11 and 22.[1,5,6] The frequency of this syndrome has been reported as 1 in 110,000 births.[2] The major anomalies are ear pits, followed by micrognathia (34%), congenital heart disease, cleft palate, seizure, preauricular tags, and microtia.[1,2,4,5]

Congenital heart disease is frequently associated with Emanuel syndrome.[2,3] Depending on the degree and type of heart disease, careful monitoring of hemodynamics is critical (eg, central venous pressure, pulmonary artery wedge pressure, and/or CO). However, insertion of central venous catheter or pulmonary artery catheter would be considered a more invasive procedure compared to the procedure of palatoplasty. In this patient, the ASD was reasonably small with predominant left to right shunt and the PS would be considered mild. However, anesthetic agents and hemodynamic responses to them may cause significant circulatory disturbances. Therefore, anesthetic management based on adequate noninvasive cardiac function monitoring was used. In this case, cardiac function was monitored by Aesculon®, a noninvasive cardiac function monitor using the electrical velocimetry method. The Aesculon® uses standard ECG surface electrodes attached side to side in a vertical direction to the patient's left middle and lower neck and to the lower thorax at the left mid-axillary line at the level of the heart and xiphoid process. The electrodes are then connected to the Aesculon® monitor. The signal quality is verified by visualization of the ECG and the impedance waveform. Cardiac functions are calculated by transformation to the ohmic equivalent of the mean aortic blood flow acceleration and HR correction.[7] Although the absolute accuracy of the Aesculon device's ability to determine cardiovascular parameters has been questioned, we used it to follow trends in hemodynamic response.

At the induction of anesthesia, the baseline value of the SV was 7.6 mL, SVV was 16%, HR was 125 bpm, and CO was 1.1 L/min. Approximately 15 minutes after induction of anesthesia, hemodynamic responses increased. This increase in cardiac function may have been due to agitation by inhalational of sevoflurane, administration of atropine, and/or movement of the head and tube while draping and head wrap. It gradually improved over 30 minutes. However, no acute cardiovascular event, such as arrhythmia or heart failure, occurred. During surgery, no other significant changes were observed in cardiac function.

Sevoflurane is a widely used volatile anesthetic, especially for inhalation induction. However, it may induce seizure-like activity, particularly in combination with hyperventilation, which may decrease cerebral blood flow. We therefore maintained $EtCO_2$ within the normal level. Since patients with Emanuel syndrome potentially have seizure disorders,[1,4] we utilized the BIS monitor. The BIS monitor was used to assess seizure activity, as has been reported, and none was observed in this case.[9] It was useful to obtain information not only about anesthetic depth but also epileptogenic activity from its transformed EEG. Propofol and isoflurane could be utilized to suppress induced seizures had they occurred.[1,8]

Airway management depends on evaluation of the abnormalities in each case, and modifications of technique might be needed to decrease the risk of airway complications.[6]

CONCLUSION

We experienced the anesthetic management of a patient with Emanuel syndrome. Attention was particularly directed at monitoring cardiac function and possible seizure activity. Airway concerns were minimized in this case due to the presence of a tracheostomy tube.

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