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Anesthetic management of a patient with partial trisomy 18

NORIKO OCHIAI, RYU OKUTANI, and YUMI YOSHIMURA

Department of Anesthesiology, Hyogo Prefectural Nishinomiya Hospital, 13-9 Rokutanji-cho, Nishinomiya, Hyogo, 662 Japan

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Introduction

Trisomy 18 (Edwards' syndrome) is an autosomal abnormality with visceral deformities and delayed mental and motor development, including heart deformity and external deformity such as small jaw, short neck, and contracted or bending fingers. The survival rate at 1 year or more after birth is reported to be about 10% [1,2]. Partial trisomy is generally mild in comparison with full trisomy 18. Thus, although it is inferred that patients with partial trisomy 18 survive longer and may have more opportunities to receive surgery in comparison with patients with full trisomy 18, there have been no reported cases of patients with partial trisomy 18. We report the anesthetic management for tonsillectomy in a patient with partial trisomy 18 who had apneic attacks while sleeping.

Case report

The patient was a female who was 5 years and 10 months old. Her height was 81 cm and her body weight was 11 kg. She was delivered naturally at 41 weeks of gestation, and the Apgar score was 7 points at 1 min and 8 points at 5 min. Because her body weight at birth was only 2370 g (small for date), she was hospitalized at our prematurity center. The patient had both eyes isolated, small jaw, a small mouth, low position of the auricle, short neck, bent and contracted fingers, and insufficient nail formation. Myoclonus-like tremors occurred frequently. Echocardiography showed a small ventricular septal defect (VSD) and a karyogram demonstrated translocations of chromosome 18. Because the parents had no chromosomal abnormalities, the patient's abnormality was considered to be due to a mutation. No abnormalities were found on the electroencephalogram and the myoclonus-like tremor gradually disappeared. The VSD appeared to have closed naturally when the patient was 2 years old. Her medical history included pneumonia at 2 years and bronchitis at 3 years of age for which she was hospitalized. She frequently caught colds.

From the age of about 3 years, nasal congestion and loud snoring during sleep became conspicuous, and she suffered repeated apneic attacks in the supine position during sleep. Apneic attacks occurred frequently and became severe from the age of 5 years. An examination at the Department of Otolaryngology at our hospital revealed hypertrophic bilateral tonsils (grade III), which were suspected to be the cause of the apneic attacks. Resection of the bilateral tonsils was scheduled. Presurgical examinations showed no abnormalities in the hematologic or urinary biochemical examinations or on electrocardiogram, and the echocardiography reconfirmed the natural closure of the VSD. The electroencephalogram did not show abnormalities such as epileptiform waves. The external deformities identified at birth were confirmed and funnel chest was also found. Her mental and motor development was that of a 1-year-old; she could only mutter and crawl.

Because the patient had a temperature (38°C) and cough with mild pharyngeal rubor on the morning prior to surgery, fluid infusions and antibiotics were administered by the peripheral venous route. Her symptoms had improved by evening and her temperature declined to 36.3°C. She had only a mild cough in the morning on the day scheduled for surgery. Accordingly, the surgery was performed as scheduled.

Address Correspondence to: R. Okutani
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Anesthetic course

The premedication consisted of 0.1% diazepam syrup (7 ml) administered orally 2 h, and an intramuscular injection of atropine sulfate (0.2 mg) given 30 min before the patient entered the operating room. In the operating room, an electrocardiograph, automatic blood pressure monitor, and pulse oximeter were set up, and oxygen (100%) and isoflurane (1%) were introduced. Endotracheal intubation was conducted with vecuronium (1 mg). Maintenance was carried out with oxygen, nitrous oxide (60%), and isoflurane (1.5%), and vecuronium was added as needed. No notable changes in rectal temperature, blood pressure, heart rate, or respiratory status were observed during surgery. Extubation was conducted after the reversal of the residual effect of vecuronium with neostigmine (0.6 mg) and atropine sulfate (0.2 mg). Because her systemic condition was stable and she promptly emerged from anesthesia, she was taken to the general ward after 30 min follow-up in the operating room.

The apneic attacks during sleep began to decrease in number soon after surgery and were markedly reduced 4 days later.

Discussion

The incidence of full trisomy 18 is 1 in every 3500–7000 births, and it leads to death at the newborn or infant stage in most cases [1]. External deformities include deformity or low set of the auricules, small jaw, eyes isolated, small neck, bent and contracted fingers, limited hip abduction, and dorsiflexion of the great toe. Severe congenital heart disease, visceral deformities such as Meckel's diverticulum and cystic kidneys, physical and mental retardation, and spasmodic attacks were also seen [2]. On the other hand, the incidence of partial trisomy 18 is very rare and only about 10 cases have been reported in the world [3]. However, in general, partial trisomy 18 is mild in comparison with full trisomy 18. Although these children have delayed mental and motor development, they sometimes survive for many years [4]. Our patient had most of the characteristics of full trisomy 18 except for severe cardiac malformation and visceral deformity.

Key points in the anesthetic management of such patients are preoperative examination for the presence or absence of congenital heart disease, difficulties in ventilation with a mask and with tracheal intubation at the time of anesthetic induction, and possibility of occurrence of malignant hyperthermia. Bailey and Ghung [5] reported their anesthetic experience using a laryngeal mask for myringotomy in a 3-year-old female with trisomy 18 with a favorable outcome. It was not mentioned whether the laryngeal mask was used because surgery was only a short-duration operation or because endotracheal intubation was difficult. On the other hand, Matsuda et al. [6] reported a 3-year-old male with trisomy 18 who was scheduled for fixation of the testicle and developed muscular rigidity after administration of succinylcholine. Endotracheal intubation became impossible [7]. The patient was managed under mask anesthesia and had a high temperature (38.4°C) and an elevated creatine phosphokinase level during surgery. As this is the only reported case of trisomy 18 in which malignant hyperthermia developed during surgery, the relationship between malignant hyperthermia and trisomy 18 is not clear.

Respiratory management including securing of the airway is important in anesthetic management in oral surgery such as resection of the tonsils. Excessive sedation by means of premedication inhibits respiration. The anatomical characteristics of small jaw and small mouth may make mask ventilation and endotracheal intubation difficult. To have prompt emergence and to prevent postoperative respiratory inhibition, an inhaled anesthetic is better than fentanyl. In the postoperative follow-up, sufficient hemostasis should be confirmed to prevent hemorrhage after extraction of the tonsils, and the systemic condition, particularly respiration should be observed carefully to prevent choking. In this case, the venous route was secured before surgery and rapid induction was possible. It was also fortunate that mask ventilation and endotracheal intubation were conducted easily. However, on the assumption that tracheal intubation may be difficult, it is necessary to conduct anesthetic induction carefully and to prepare a laryngeal mask and bronchofiberscope.

Although this patient showed pyrexia and inflammatory symptoms of the upper airway 1 day prior to surgery, administration of antibiotics had relieved these symptoms by the following morning. Considering that the patient had repeated colds and that the pyrexia might have been due to the hypertrophic tonsils, we performed surgery as scheduled.

The relationship between trisomy 18 and apneic attack during sleep was unknown. Grade III tonsillar hypertrophy was present in our patient. We were not able to determine whether the apneic attack was central, peripheral, or mixed type. However, the tonsillar hypertrophy might have exacerbated the clinical symptoms. Therefore, surgery was indicated from the viewpoint of differential diagnosis. Although the apneic attacks during sleep did not disappear soon after surgery, the frequency of the attacks was markedly reduced 4 days postoperatively. Thus, the main cause for apneic attack was assumed to be tonsillar hyertrophy, a peripheral factor. The patient had no complications and was discharged from the hospital 10 days after operation.

In summary, we experienced anesthetic management for tonsillectomy of girl with a partial trisomy 18. The anesthesia was safely completed without difficulty in tracheal intubation or occurrence of malignant hyperthemia.

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