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Case Report

Airway Management of Patient with Smith-Lemli-Opitz Syndrome

Ahmet Küçük[1], Harun Aydoğan[1], Mahmut Alp Karahan[1], Hasan Hüsnü Yüce[1], Nuray Altay[1], İsmail İynen[2]

Abstract

Smith–Lemli–Opitz syndrome (SLOS) is a rare autosomal recessive syndrome presenting with multiple congenital anomalies and mental retardation associated with low plasma cholesterol levels. Two forms have been recognized based on clinical course and severity: the classical SLOS (type I) and the lethal acrodysgenital syndrome (type II). SLOS type I clinical symptoms are less severe, and life expectancy is longer. SLOS type II is also accompanied by multiple congenital anomalies, and the prognosis is poor, which is lost within weeks. Micrognathia, cleft palate, including large and abnormal stiff tongue, and typical dysmorphic facial features are some of the major causes for the difficult airway in this syndrome. In this case report, we discussed the airway management of a child with SLOS type I who underwent bilateral ear tube insertion.

Keywords: Smith–Lemli–Opitz syndrome, laryngeal masks, airway management

Introduction

Smith–Lemli–Opitz syndrome (SLOS) is a multiple malformation syndrome, in which a congenital error of sterol synthesis was first detected in human beings. This syndrome, which demonstrates autosomal recessive inheritance, was first defined by Smith, Lemli, and Opitz in 1964 (1). Decreased plasma cholesterol levels associated with a defect in 3-beta-hydroxycholesterol 7-reductase, which converts 7-dehydrocholesterol (7-DHC) to cholesterol in cholesterol biosynthesis, and increased plasma concentration of 7-DHC are observed. The incidence of SLOS is stated to be 1 in 15,000–40,000 among Caucasians. While SLOS is more frequently seen in Northern and Central Europe, it is less frequent in Asia and Africa (2). There is an inverse correlation between the level of serum cholesterol and clinical severity of the disease. It is classified into 2 groups as type I (classical form) and type II (acrodysgenital syndrome) according to the clinical course and prognosis. In type 1 SLOS, clinical symptoms are milder and lifetime is longer. Moreover, isolated cleft palate, bifid uvula, syndactyly of fingers 2 and 3, cataract, growth retardation, and behavioral disorders can be seen. On the other hand, in type II SLOS, congenital anomalies occur with ambiguous genitalia. Most of the time, its prognosis is poor and patients are lost within weeks (3). Considering congenital anomalies, anesthesia is needed for diagnostic interventions and corrective surgeries in SLOS patients. The most common craniofacial features of SLOS include microcephaly, bitemporal narrowing, ptosis, short helix-structured nose, micrognathia, epicanthal folds, high-arched and narrow hard palate, sublingual tissue excess, bifid uvula, and cleft palate. With these features, SLOS patients are candidates of difficult airway (4). In this case report, airway management was discussed in a 4-year-old patient diagnosed with type I SLOS and in whom it was planned to insert bilateral ear tubes.

Case Report

In a 4-year-old, 16 kg, and 95-cm male patient with the diagnosis of type I SLOS, surgeons in the ear–nose–throat (ENT) clinic planned to insert bilateral ear tubes under general anesthesia. In the preoperative anesthesia examination of the patient, it was found that he had a dysmorphic facial appearance (Figure 1a, b), mental and motor retardation, bifid uvula (Figure 2a), and typical syndactyly of the 2nd and 3rd toes in the lower extremities (Figure 2b). His Mallampati score was 2, and there was no restriction in his neck flexion and extension. In the laboratory analyses, the level of serum cholesterol was 95 mg/dL (112–200 mg/dL), and other biochemical analyses results and complete blood count were within normal intervals. As a result of pediatric consultation, no cardiac and respiratory findings were found. Written informed consent form was obtained from his parents. The patient was taken to the operating room and was pre-oxygenized with 100% O_2 for 3 min. For sedation, 1 mg of intravenous midazolam was applied. The patient was monitored with standard electrocardiography; non-invasive blood pressure (NIBP), peripheral oxygen saturation (SpO_2), and end-tidal CO_2 ($EtCO_2$) measurements; and using a heat probe. His SpO_2 was 100%, heart rate was 130 beats/min, NIBP was 90/40 mmHg, and body temperature was 36.7°C. Following pre-oxygenation, the patient was intravenously administered 1 mg/kg of lidocaine, 0.5 µg/kg of remifentanil, and 2

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[1]Department of Anesthesiology and Reanimation, Harran University Faculty of Medicine, Şanlıurfa, Türkiye
[2]Department of Otorhinolaryngology, Harran University Faculty of Medicine, Şanlıurfa, Türkiye

Address for Correspondence:
Mahmut Alp Karahan
E-mail: mahmutalp_k@yahoo.com

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Figure 1. a, b. (a) SLOS dysmorphic facial appearance, (b) SLOS LMA application
SLOS: Smith–Lemli–Opitz syndrome

Figure 2. a, b. (a) SLOS bifid uvula, (b) SLOS syndactyly of the 2nd and 3rd toes
SLOS: Smith–Lemli–Opitz syndrome

mg/kg of propofol for anesthesia induction. He was made to inhale 100% oxygen, and a $2_{1/2}$-sized laryngeal mask airway (LMA) was properly placed (Figure 1b). Minute ventilation was adjusted according to the $EtCO_2$ value. The maintenance of anesthesia was provided with 2% sevoflurane, 50% oxygen, and 50% dry air. The surgical procedure lasted for approximately 20 min. LMA was removed when spontaneous respiration and protective reflex were observed in the patient who did not have any complication. Because his spontaneous respiration was adequate and hemodynamic parameters were normal, he was taken into the post-anesthesia care unit. Then, the patient, whose vital findings were stable, was transferred to the ENT clinic. With no development of any complication, he was discharged from the clinic on the postoperative 2nd day.

Discussion

Smith–Lemli–Opitz syndrome is a rarely seen syndrome that demonstrates autosomal recessive inheritance accompanied by the biochemical impairment of cholesterol synthesis, and the cumulative effect of toxic cholesterol precursors and cholesterol deficiency explains variable phenotypes of this syndrome. There is a report of an inverse correlation between the level of serum cholesterol and clinical picture of the disease (3). In type I SLOS patients, the spectrum of physical findings is wide and includes cataract, syndactyly of fingers 2 and 3, bifid uvula, and developmental and behavioral disorders. On the other hand, in type II SLOS, non-vital genital and multiple congenital anomalies are seen in the neonatal period (5). Airway management is important for anesthesia in SLOS patients. Typical craniofacial defects such as micrognathia, cleft palate, and small and abnormal hard tongues can cause difficulty in laryngoscopy and intubation in SLOS patients (6). Our patient diagnosed with type I SLOS had bifid uvula, syndactyly of the 2nd and 3rd toes, and mental and motor development retardation.

Smith–Lemli–Opitz syndrome patients are considered as a potential patient group for difficult endotracheal intubation. In these patients, palate anomalies such as micrognathia (67–10%0), tongue anomalies (12–63%), cleft palate, and high-arched palate (37–68%) are among the main causes for laryngoscopy and intubation difficulty (7). There is a limited number of studies reporting airway management in SLOS patients. In literature, it is found that alternative methods are used for the approach to and management of airways in such patients (4, 6).

In a 10-year-old and 15-kg SLOS patient who underwent esophagoscopy under general anesthesia for removing a foreign body in the esophagus, micrognathia, microcephaly, and cleft palate were detected with a typical dysmorphic face. The patient was considered to be Cormack–Lehane score IV in the laryngoscopic evaluation, and intubation was successfully performed with the help of an Airtraq optic laryngoscope (8). In a study on 20 surgical interventions performed on 14 SLOS patients between 2000 and 2002, anesthesia administration and airway management were investigated. It was reported that 5 of 14 patients in the series had difficult airways, and successful intubation processes with fiberoptic bronchoscopy were emphasized (9).

Characteristic facial appearance is a sign of difficult intubation, which requires necessary precautions to be taken for providing airway management. In this patient group, it is essential to provide an airway with LMAs in suitable operations. At present, LMA is commonly used, and it is an alternative and successful choice for patients with difficult airways, including those with SLOS. In a 5-day-old and 2.4-kg SLOS patient in whom pylorotomy was to be performed, surgeons planned to perform awake direct laryngoscopy. However, because it failed, fiberoptic intubation was attempted. Because of failure again, LMA was placed, and the operation was completed (10). In another study, it was reported that standard intubation techniques were unsuccessful in a premature newborn with SLOS and that ventilation could be provided with a classical LMA (11). In an SLOS patient accompanied by thrombocytopenia, a difficult airway was encountered due to factors such as dysmorphic face, micrognathia, and abnormal tongue, and airway management was succeeded using LMA without any complication (12). In our case, LMA was preferred for airway management because the surgical intervention would be short and the patient had bifid uvula and it was successfully applied.

Conclusion

In SLOS patients, the preoperative evaluation of airway with regard to difficult airway should be perfectly performed because of anatomic anomalies. It is suggested that LMA, which provides positive pressure ventilation, continuous positive airway pressure, and spontaneous respiration safely, is a good alternative for airway management.

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