OLGU SUNUMU CASE REPORT

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Our Outpatient Anesthesia Management in a Pediatric Case with Cornelia de Lange Syndrome

Cornelia de Lange Sendromlu Pediatrik Olguda Günübirlik Anestezi Yönetimimiz

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ABSTRACT Cornelia de Lange syndrome (CdLS) is a rare congenital syndrome accompanied by significant craniofacial, cardiovascular, gastrointestinal, and musculoskeletal anomalies with dysmorphic facial appearance. As many systems are affected in these patients, anesthesia management can become complex. An 8-year-old, 17 kg, a 31-month-old girl diagnosed with CdLS due to developmental delay applied for cranial and whole spinal magnetic resonance imaging (MRI). The Mallampati score was III in the preoperative examination, the mouth opening was narrow, and the risk of difficult intubation was high. We have prepared all necessary materials following the guidelines. MRI was successfully performed while maintaining the patient's spontaneous breathing under oxygen support. This case report is aimed to raise awareness for this syndrome, where difficult airway and intubation are expected in specialized units such as non-operating room anesthesia applications, and to review it in the light of the literature.

Keywords: De Lange syndrome; anesthesia; outpatients

ÖZET Cornelia de Lange sendromu (CdLS); kraniyofasiyal, kardiyovasküler, gastrointestinal, kas-iskelet sisteminin majör anomalileri ile dismorfik yüz görüntüsünün eşlik ettiği nadir konjenital bir sendromdur. Bu hastalarda birçok sistem etkilendiği için anestezi yönetimi karmaşık hâle gelebilir. Sekiz yaşında, 17 kg, 31. ayında gelişme geriliği nedeniyle CdLS tanısı alan kız çocuk, kraniyal ve tüm spinal manyetik rezonans görüntüleme (MRG) için getirildi. Preoperatif muayenesinde Mallampati skoru III, ağız açıklığı dar, zor entübasyon riski yüksekti. Kılavuzlara uygun olarak gerekli tüm malzemeler hazır edildi. Yaklaşık 45 dk. süren MRG, oksijen desteği altında hastanın spontan solunumu korunarak başarıyla gerçekleştirildi. Bu olgu sunumunda, günübirlik anestezi uygulanan özellikli ünitelerde, zor havayolu yönetimi beklenen bu sendrom için farkındalık yaratmak ve literatür eşliğinde gözden geçirmek amaçlanmıştır.

Anahtar Kelimeler: De Lange sendromu; anestezi; ayaktan hastalar

Cornelia de Lange syndrome (CdLS) is a rare congenital syndrome characterized by motor and mental retardation, short neck, micrognathia, microcephaly, high palate, hypertrichosis, major anomalies of the cardiac, gastrointestinal, and musculoskeletal systems, and dysmorphic facial appearance.[1] The most common chromosomal abnormalities are partial trisomy in the long arm of the 3rd chromosome (3q26.1) or monosomy in the long arm of the 9th chromosome.[2] The prevalence of CdLS varies between 1/10,000 and 1/50,000, and the probability of recurrence in subsequent pregnancies is 2-5%.[3]

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Congenital heart diseases are seen in 25-30% of the patients, epilepsy in 20%, and gastroesophageal reflux in more than 85-90%.[4] Various problems, especially difficult airways, should be expected due to different deformities and congenital anomalies in the anesthesia management of these patients. There are limited studies in the literature on the anesthetic management of patients with this syndrome.

This case report is aimed to raise awareness for this syndrome, where difficult airway and intubation are expected in specialized units such as nonoperating room anesthesia applications, and to review it in the light of the literature.

CASE REPORT

The patient, an 8-year-old girl with a height of 17 kg and 89 cm, was admitted to our magnetic resonance imaging (MRI) center for cranial and whole spinal MRI. She was diagnosed with CdLS (peripheral blood-DNA analysis: 46XX without NIPBL and SMC1A gene mutation) due to a first-degree consanguineous marriage by the pediatric neurology and medical genetics physicians she applied to when she was 31 months old. He had a history of upper gastrointestinal system endoscopy with diagnostic sedation at three years old. He had no known history of cardiac disease and no drug use.

The patient had typical dysmorphism findings such as prominent eyelashes, flattened nasal root, micrognathia, increased facial hair, long anterior teeth, short neck, the length difference between the thumbs, and 2-3 syndactyly of the right foot (Figure 1). She was admitted with 6-hour fasting on the day of the appointment, after routine preoperative anesthesia examination, laboratory examinations, and written voluntary patient consent. The preoperative assessment evaluated Mallampati score III, narrow mouth opening, and high risk of difficult intubation. MRI compatible anesthesia machine, monitor, laryngoscope, ambu-oxygen system, difficult intubation stylet, endotracheal tubes of different sizes, and laryngeal and face masks were checked. Following standard monitoring, midazolam 1 mg IV was administered, and the MRI device was placed supine. Heart rate was 125 130 beats min-1, blood pressure was 100/70 mmHg, and SpO_2 was 99%.

FIGURE 1: Our patient has prominent eyelashes, flattened nasal roots, thin lips, downward-facing mouth corners, micrognathia, increased facial hair, widely spaced teeth, and short stature.

Although ketamine propofol sedation was considered to preserve spontaneous respiration, it was abandoned because of the possibility of exacerbating tachycardia. 2-4 L min-1 oxygen was started with a nasal cannula. Thiopental 4 mg kg-1 slow IV the push was made. 1 mg kg-1 IV intermittently while maintaining spontaneous respiration. Three additional doses were administered. The MRI procedure took approximately 45 minutes. Nausea, vomiting, apnea, desaturation, and hemodynamic instability were not observed in the patient followed up in the review.

The patient and her mother were informed about the case report. The patient's mother obtained written and verbal consent to present the case and facial photograph. The ethics committee admission was not made because it was a case report.

DISCUSSION

The difficult airway is emphasized in almost all studies in which anesthesia management of patients with CdLS is included. Patients have a typical facial appearance, as pathologies such as cleft palate, microcephaly, long eyelashes, hirsutism, flattened nasal root, long nasal philtrum, micrognathia, short neck, mandible anomalies, choanal atresia, craniofacial or orofacial deformities, and high palate can be encountered the potential risk for difficult

airway and intubation.1-4 A classical laryngeal mask, Fastrack laryngeal mask, or fiberscope-assisted intubation in pediatric patients has been suggested as an effective alternative to tracheal intubation. Defects in tooth structure and mental erosions secondary to gastroesophageal reflux may complicate laryngoscopy. Patients are likely to develop gastroesophageal reflux due to poor esophageal motility and hiatus hernia, and there is an increased risk of aspiration pneumonia.[5,6]

Yokoyama et al. failed orotracheal intubation in a young patient with limited mouth opening but succeeded in blind nasotracheal intubation. They stated that nasotracheal intubation is more advantageous in such patients by maintaining spontaneous breathing with the help of inhalation anesthetics.[7]

Since organ immaturity is typical, a small endotracheal tube is recommended, especially in children with laryngeal hypoplasia.[8] Furuya et al. reported that they performed endotracheal intubation without using neuromuscular blockers in a 22-month-old boy with micrognathia and a short neck who underwent palatoplasty. They used dexmedetomidine and sevoflurane for induction and provided laryngeal reflex inhibition with propofol.[9]

Park et al. reported that the video laryngoscope was helpful in intubation in a typical child with a short neck, micrognathia, and restricted mouth opening.[10] Hirai et al. reported that they could provide endotracheal intubation with the help of a fiberoptic bronchoscope in a 1-year-old baby.[11]

It has been stated that congenital heart diseases can cause hypoxic attacks and difficult airways. Nakajima et al. planned dental treatment under general anesthesia in a 21-year-old CdLS patient with tetralogy of Fallot. They reported successful endotracheal intubation with fiberoptic bronchoscopy while maintaining spontaneous respiration with sevoflurane and nitrous oxide.[12]

Moschini et al. in their retrospective study of 42 patients with CdLS, stated that the problems were similar and required one patient's use of a special laryngoscope (McCoy blade).[13] Torres et al. stated that endotracheal intubation was performed with the help of a fiberoptic bronchoscope through the laryngeal mask airway placed after induction with inhalation anesthetics. In cases where it is thought that difficult intubation may occur, it was emphasized to plan anesthesia induction by maintaining spontaneous breathing.[14]

Acute pneumonia or bronchitis is the most common cause of death in children with CdLS. In these patients, endotracheal intubation should be done carefully when necessary, and the depth of anesthesia should be controlled. Since patients are susceptible to respiratory tract infections and the airway is considered irritable, anesthesia and operation time should be kept short. Care should be taken with anesthetics (e.g., enflurane, ketamine, droperidol) as they may cause convulsions in CdLS. Even if a motor seizure is not detected in a patient with epileptic EEG waves, care should be taken to prevent it, and these agents should be avoided. Hyperventilation and high body temperature have also been reported to cause convulsions.[15]

In the preoperative evaluation of our patient, Mallampati III was evaluated as having a narrow mouth opening and a high risk of difficult intubation. Our patient had no history of epilepsy or seizures. Although ketamine-propofol was considered for the patient's sedation, whose heart rate was between 125-130 beats min-1 after standard anesthesia monitoring, it was abandoned because it might increase tachycardia.

Thiopental was chosen because it is thought to protect spontaneous respiration. The MRI procedure takes approximately 45 minutes. It maintained spontaneous breathing while it was in progress and completed the operation successfully without intubation.

In conclusion, CdLS should be considered in these patients with typical facial morphology. It is impossible to establish standard anesthesia management in patients with CdLS. Individual planning should be done. Difficult airway and difficult intubation should be expected, especially in outpatient anesthesia applications, and spontaneous breathing should be avoided. When endotracheal intubation is required, all preparations should be made meticulously.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Kadir Arslan, Aykan Gülleroğlu;* ***Design:*** *Kadir Arslan;* ***Control/Supervision:*** *Kadir Arslan, Aykan Gülleroğlu;* ***Data Collection and/or Processing:*** *Kadir Arslan;* ***Analysis and/or Interpretation:*** *Kadir Arslan, Aykan Gülleroğlu;* ***Literature Review:*** *Kadir Arslan;* ***Writing the Article:*** *Kadir Arslan;* ***Critical Review:*** *Kadir Arslan, Aykan Gülleroğlu;* ***References and Fundings:*** *Kadir Arslan.*

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