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Anesthesia Management in Cornelia de Lange Syndrome

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Case Report

Abstract

Cornelia de Lange Syndrome (CdLS) is a genetic disorder characterized by a variety of clinical features, including motor and intellectual disabilities, short neck, microcephaly, micrognathia, high-arched palate, distinctive facial features, and limb anomalies. In this case presentation, we discuss a 1.5-year-old child with CdLS who was scheduled for bilateral undescended testis surgery. We aim to highlight the considerations and challenges in selecting the appropriate anesthesia procedure for a patient with CdLS, particularly taking into account the associated abnormalities and potential complications.

Keywords:
Cornelia de Lange Syndrome;
anesthesia;
undescended testis surgery

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Introduction

Cornelia de Lange Syndrome (CdLS), also known as Brachmann–de Lange Syndrome, is a rare genetic disorder with an estimated frequency of 1 in 10,000 to 1 in 50,000 live births. The syndrome presents with motor and intellectual disabilities, short neck, microcephaly, micrognathia, high-arched palate, a characteristic facial appearance, and limb abnormalities [1]. In some cases, partial trisomy of the long arm of chromosome 3 (3q26.1) or monosomy of the long arm of chromosome 9 are the most frequently described chromosomal anomalies [2,3].

Anesthesia management in CdLS presents several challenges, including difficult intubation, aspiration risk, behavioral disorders, severe scoliosis, recurrent respiratory infections due to impaired lung function, difficult venous access, and the potential risk of malignant hyperthermia [3]. Common problems during general anesthesia include gastroesophageal reflux, aspiration, and hyperthermia [4]. This case report presents the anesthesia management of a CdLS patient undergoing a urogenital operation.

Case Report

A 1.5-year-old, 10 kg patient with CdLS was scheduled for bilateral undescended testis surgery. On physical examination, the patient exhibited microbrachycephaly with a small face, thick arched eyebrows, long eyelashes, anteverted nostrils with a depressed nasal bridge, long philtrum, thin downward-curved upper lip, missing teeth, low-set

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ears, micrognathia, and a short extended neck (Figures 1 and 2]. The patient had severe growth and speech delay, along with intellectual disability. Routine biochemical investigations were within normal limits, and echocardiography revealed no abnormalities.

Various sizes of face masks, oral and nasopharyngeal airways, endotracheal tubes, LMA, and I-gel devices were prepared. Anesthesia induction was initiated with sevoflurane, followed by propofol (3 mg/kg), fentanyl (2 µg/kg), and remifentanil (0.25 µg/kg). After adequate face mask ventilation, the airway was secured using an I-gel device. Anesthesia was maintained with propofol infusion at 75 µg/kg/min and remifentanil infusion at 0.2 µg/kg/min using total intravenous anesthesia (TIVA).

The surgery lasted approximately 45 minutes. Postoperatively, there was a slight delay in awakening. Once normal respiratory function, muscle tone, and reflexes were restored, the patient was extubated and transferred to the post-anesthesia care unit (PACU) for monitoring.

Discussion

The etiology of CdLS is not fully understood. Microscopic examinations have shown a reduction in oligodendroglial cells and myelin defects in transverse fibers [5]. A complete absence of pregnancy-associated plasma protein A (PAPPA) has also been associated with CdLS. Most cases are sporadic; however, in cases with a family history or consanguinity, a 3q26.3 chromosomal defect has been observed [6]. Genetic inheritance is characterized by low-penetrance autosomal dominant or autosomal recessive patterns.

Due to the high frequency of complications, careful pre-anesthetic evaluation and airway examination are essential [7]. Difficult airway is expected due to craniofacial and orofacial deformities such as short neck, small mouth, and high-arched palate. Dental abnormalities and erosion due to gastric reflux may further complicate laryngoscopic procedures [8].

Supraglottic airway devices (SGAs) are essential tools for managing difficult pediatric airways. They provide adequate airway patency, oxygenation, and ventilation, particularly in patients with anatomical challenges. In our patient, craniofacial anomalies such as microcephaly, micrognathia, macroglossia, a short hyperextended neck, and widely spaced or missing incisors indicated a difficult airway. Therefore, an I-gel device was chosen, which provided secure airway management without the need for neuromuscular blockade.

Although some CdLS cases have been speculated to carry a risk of malignant hyperthermia (MH), no definitive evidence confirms a direct association. As a precautionary measure, we avoided both depolarizing and non-depolarizing neuromuscular blocking agents. Volatile anesthetics such as sevoflurane, despite their known potential to trigger MH in susceptible individuals, have been reported to be used safely in CdLS without MH-related complications [9–11]. In our case, sevoflurane was selected for inhalational induction due to its rapid onset, low airway irritability, and established safety profile in pediatric patients. MH preparedness protocols, including vigilant intraoperative monitoring and emergency readiness, were implemented. The perioperative period was uneventful, and no signs of MH were observed.

Conclusion

Anesthesia management in Cornelia de Lange Syndrome is challenging due to the multisystemic nature of the disease. Successful management requires careful pre-anesthetic assessment, appropriate preparation, and meticulous postoperative monitoring.

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Ethics

Informed Consent
The patient provided consent for the clinical information pertaining to the case to be published in a medical journal.

Author Contributions
Surgical and Medical Practices – E.C., F.C.; Concept – E.C., F.C.; Design – E.C., F.C.; Data Collection and/or Processing – E.C., F.C.; Analysis and/or Interpretation – E.C., F.C.; Literature Review – E.C., F.C.; Writing – E.C., F.C.

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