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Anesthetic Management for Escobar Syndrome: A Case Report

Escobar Sendromunda Anestezi Yönetimi: Olgu Sunumu

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Abstract / Öz

Escobar syndrome is a genetic disorder that causes orthopedic and respiratory malformations. Patients with Escobar syndrome are important by anesthesia management due to various airway difficulties and the presence of malignant hyperthermia. In this case, we administered I-Gel for the anesthetic management of a 4-month-old child undergoing various orthopedic surgeries in her lower extremity.

Keywords: Escobar syndrome, airway management, malignant hyperthermia, supraglottic airway devices

Escobar sendromu, ortopedik ve solunum bozukluklarını içeren genetik bir hastalıktır. Bu sendromlu hastalar, çeşitli hava yolu zorlukları ve maling hipertermi nedenleriyle anestezi yönetiminin karakteristik özelliklerini taşırlar. Bu olguda, alt ekstremitede çeşitli ortopedik operasyonlar geçirecek 4 aylık bir çocuğun I-Gel ile anestezi yönetimini sunduk.

Anahtar Kelimeler: Escobar sendromu, havayolu yönetimi, malign hipertermi, supraglottik havayolu cihazları

Introduction

Escobar syndrome is a rare autosomal recessive disorder, which is also called "multiple pterygium syndrome" (1). This disease is characterized by axillary, antecubital, popliteal, digital, and intercrural joint flexion contractures; growth retardation; pterygium in the eyes; decreased lung capacity; genital abnormalities; and vertebral anomalies (2-6). Furthermore, most Escobar syndrome patients have airway problems such as increased kyphosis, cervical fusion with excessive cervical lordosis, limited movement due to neck flexion contractures, cleft palate, ankyloglossia, micrognathia, and a restricted mouth opening. As a result, it is important for Escobar syndrome patients to be aware of airway management control (3, 7-8).

In this case report, we present the anesthetic evaluation of a child diagnosed with Escobar syndrome undergoing pediatric orthopedic surgery.

Case Report

A 4-month-old girl weighing 10 kg was admitted for open reduction of right hip dislocation, right pes equinovarus, achilloplasty, vertical talus on the left side, tibialis anterior release, and left ankle joint posterior capsular release. Informed consent was obtained from the patients' relatives. Preoperatively, the mouth, jaw, and neck joint clearance, respiratory and cardiovascular systems, and laboratory analyses of the patient were examined. Although the preoperative cardiac examination (including echocardiography and electrocardiography), routine hematological test results, and posterioranterior chest x-ray were within normal ranges, the mouth, jaw, and neck joint clearance were limited. The patient was taken to the operating table without premedication, and heart rate, temperature, non-invasive blood pressure, and peripheral oxygen saturation were monitored. The patient was taken to the area where the IV catheter was to be inserted. In this syndrome, because of the increased risk of malignant hyperthermia, patient's anesthetic management was performed with 0.01 mg of IV atropine (Atropin sulfat®, Galen, Tekirdag, Turkey) 10 mcg of fentanyl (Talinat® 0.5 mg, VEM, Istanbul, Turkey), and 30 mg of propofol (Propofol® %1 Fresenius-kabi, Hamburg, Germany) (Figure 1, Figure 2). The patient's spontaneous respiration was protected, and ventilation was achieved with a face mask. As the patient had no difficulty with mask respiration after sufficient anesthesia was provided, the airway was maintained with I-Gel supraglottic airway no: 2 (I-Gel®, Intersurgical, Berkshire, UK). Anesthetic management was provided with 50% air, 50% oxygen, and with an infusion of propofol at a rate of 50–150 mcg.kg^{-1}min^{-1}. During surgery, the patient's end-tidal carbon dioxide value, body temperature, and urine output were monitored using the urine catheter. Hemodynamic parameters and vital findings remained stable during the surgery, which lasted 2 h. At the end of the surgery, anesthesia was discontinued, and 100% oxygen was given to

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Figure 1. Preoperative photograph of a child with bilateral ptosis and facio-cranial dysmorphism

Figure 2. Preoperative monitored child with Escobar Syndrome, joint contractures (axilla, elbow, hip, knee, ankle, and neck) and limitation of movement

the patient. Superficial reflexes returned after 7 min, and I-Gel was removed following full consciousness. The patient was transported to a post-anesthesia care unit, and postoperative pain treatment with 20 mg. kg^{-1} of paracetamol (Paracetamol® ped syrup, Istanbul, Turkey) was rectally administered. After her vital signs were observed for 30 min, the patient was transported to an orthopedic surgery ward without any issues.

Discussion

Escobar syndrome is characterized by developmental retardation, dysmorphic facial appearance, pterygium present in multiple areas and akinesia. Other joint pathologies include flexion contracture, abnormal vertebral fusion, thoracic deformities, rocker bottom foot, syndactyly, camptodactyly, and dysmorphic facial appearance (9).

This syndrome's characteristics, such as cardiac and respiratory problems, kyphoscoliosis, decreased mouth opening, limited neck flexion, micrognathia, cleft palate, pterygium colli, and vertebral abnormalities, need to be considered for safe and effective anesthetic management. The possibility of airway difficulty must be anticipated during the preoperative anesthetic examination.

In this case, bilateral ptosis, dysmorphic facial appearance, and antecubital, cervical, and popliteal flexion contracture were present, and an orthopedic surgery was performed to correct these pathologies.

Anesthetic management in Escobar syndrome patients involves the assessment of the management of difficult airways and the discussion of various options. Kuzma et al. (10) concluded that awake fiberoptic-guided intubation following a failed airway in a child with Escobar syndrome provided a safe airway (with laryngeal mask airway as a substitute if endotracheal intubation was replaced). Video laryngoscopy is an alluring alternative as it provides a high-resolution view of the pediatric patient's difficult airway (11).

In this case, the patient's cleft palate became an airway problem. However, because we used I-Gel for airway management, we did not encounter any problem with the airway during the surgery.

The etiology of Escobar syndrome is unknown. However, it has been suggested that mutations within the gamma subunit of the cholinergic receptor nicotinic gamma gene of the acetylcholine receptor (AChR), which has a role in the muscle relaxant effect, is responsible for muscle contractures observed in patients with this disorder (12). Complete or severe functional disruption of fetal AChR causes lethal multiple pterygium syndrome, whereas milder alterations result in fetal hypokinesia with congenital contractures or a myasthenic syndrome later in life (13). The increased frequency of hyperthermia is a condition that must be taken into consideration in determining the anesthetic management (14). We preferred to administer IV anesthesia to the patient due to the increased risk of malignant hyperthermia. Furthermore, we had the necessary medications on hand for an emergency treatment of malignant hyperthermia. Escobar syndrome proves fatal for approximately 6% of patients due to respiratory problems such as pneumonia, dyspnea, or apnea attacks; patients may develop a restrictive thorax and secondary kyphoscoliosis during the first year. For these patients, early and effective physical therapy is important for preserving joint mobility (9).

Conclusion

Every anesthesiologist anesthetizing a child with Escobar syndrome should be aware of the presence of cardiovascular, respiratory, and orthopedic anomalies, as well as potential difficulties involving airway management or malignant hyperthermia. The presence of the difficult airway is always expected in all Escobar syndrome cases, regardless of physical or radiological findings, and multiple airway management plans must be made available. Although genetic syndromes pose to be a unique challenge for pediatric anesthesiologists, this case has been managed without any resulting issues.

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