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Anesthesia Management Of Patient With 49XXXXY Syndrome: Case Report

49XXXXY Sendromunda Anestezi Yönetimi: Olgu Sunumu
Anesteziyoloji ve Reanimasyon

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Özet

49XXXXY sendromu, yüz, genital, kardiyak ve iskelet kas sistemi malformasyonları ile karakterize bir hastalıktır. Yüz anormalliklerine bağlı zor havayolu ile karşılaşılabilir. Kardiyak malformasyonlar ise anestezi yönetimini komplike hale getirebilir. Sendromun tipik özellikleri arasında kas iskelet komponenti bulunmakta ve bu durum özellikle dikkat gerektirmektedir. Bu makalede, 49XXXXY sendromlu çocuk hastada başarılı genel anestezi yönetimimizi paylaşmayı amaçladık. Perioperatif dönemde herhangi bir komplikasyonla karşılaşmadık. Bu olgularda, uygun şartlar sağlandığı takdirde genel anestezinin güvenle uygulanabileceğini düşünmekteyiz.

Anahtar kelimeler: *49XXXXY Sendromu*, *Anestezi*

Abstract

49XXXXY syndrome is a disease characterized by facial, genital, cardiac and skeletal musculature malformations. There may be a difficult airway due to facial abnormalities. Cardiac malformations may complicate anesthesia management. Typical features of the syndrome include the musculoskeletal component, which requires attention. In this article, we aimed to share our successful general anesthesia management in a pediatric patient with 49XXXXY syndrome. We did not any complications in the perioperative period. We think that general anesthesia can be safely applied in these cases, provided that appropriate conditions.

Keywords: *49XXXXY Syndrome*, *Anesthesia*

Introduction

49XXXXY syndrome, first described in 1960 by Fraccaro et al..[1]It is extremely rare with a prevelance of 1/85000 to 1/100000 in live male births. Mental retardation, facial dysmorphism (ocular hypertelorism, upslanting palpebral fissures and flattened nasal bridge), genital anomalies, cardiac, skeletal and muscular system malformations are the main characteristics of this syndrome.[2,3]Hyperactivity, temper tantrums, shyness and antisocial behaviours were also reported in some patients.[4-6]

Up to our knowledge there are no reports on anesthesia experience in patients with 49XXXXY syndrome. Herein, we describe anesthesia management in a pediatric patient with 49XXXXY syndrome.

Case Report

A 5-year old patient with 49XXXXY syndrome was admitted to our hospital for hypospadias surgery. The patient was the second child with a birthweight of 1800 grams at 32 weeks by caesarean section to non-consanguineoushealthy parents. His mother had a history of two miscarriages and his brother was diagnosed with type 1 diabetes mellitus. His prenatal history was significant for oligohydramnios. 1 minute APGAR score was 8. Early postnatal assessment revealed dysmorphic facial features and ambiguous genitalia. The patient was evaluated for possible chromosomal abnormalities. Karyotype analysis revealed 49XXXXY karyotype. Echocardiography showed ASD that later closed spontaneously at age one. psychomotor evaluation was remarkable for delayed motor and mental development along with speech delay. He started walking at age three

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but still could not speak a word. He has been followed with radioulnar synositosis and hypothyrodism.

Preoperative physical examination showed hypospadias, dysmorphic features and delayed motor and language development. Laboratory test results were unremarkable.The patient was taken to the operation room and monitored using electrocardiogram (ECG), pulse oximetry (SpO2), non-invasive blood pressure meter (NIBP) and neuromuscular transmission (NMT) monitor. Since the patient had micrognathia (Figure 1), video laryngoscope was prepared for possible difficult airway management.

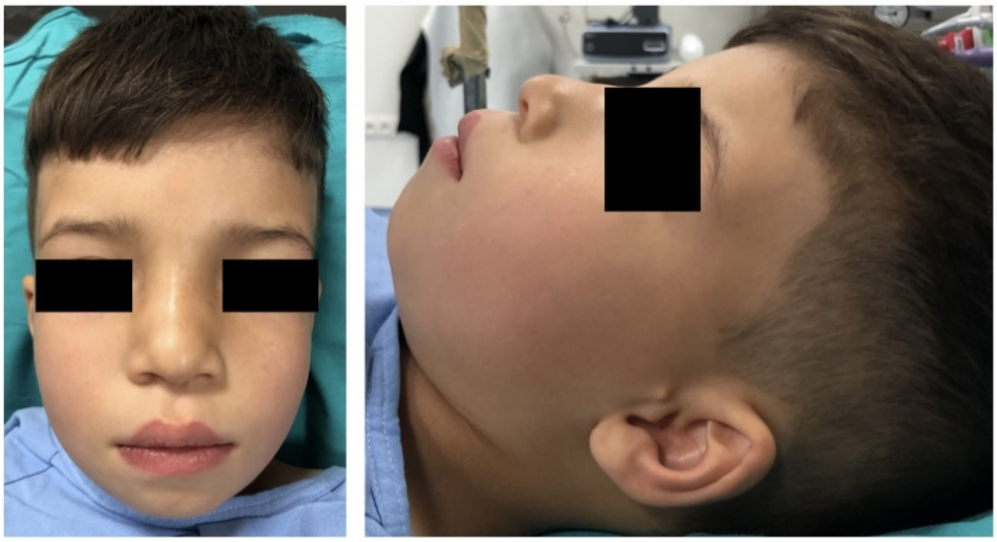

Figure 1
Video laryngoscope was prepared for possible difficult airway management

Early monitoring of the patient showed heart rate 102/min, SpO2 98% and NIBP 110/60 mmHg. Anesthesia induction was initiated. Premedication with midazolam (0.02 mg/kg) followed by propofol (2.5 mg/kg) after preoxygenation. Effective mask ventilation was easily applied. Rocuronium (0.5 mg/kg) was administered then the patient was properly intubated (TOF:0). Cormack-Lehane grade was 2. For maintenance of general anesthesia 9 mg/kg/h propofol and 0.2 mcg/kg/h remifentanil infusions were initiated. The patient developed urticarial lesions that resolved after treatment with methylprednisolone (1 mg/kg) and diphenhydramine (1 mg/kg). After 2 hours and 20 minutes surgery, the patient was successfully extubated (TOF:70). Patient vital signs were stable (HR 92/min, SpO2 100%, NIBP 92/54 mmHg). The patient developed wheezing in postoperative recovery room which resolved after steam inhalation. The patient was safely discharged from the recovery room to the ward (Modified Aldrete Score 9). On postoperative day 5, the patient was discharged from the hospital.

Case Discussion

General anesthesia can be safely administered in patients with 49XXXXY syndrome, when appropriate conditions are provided.

Facial dysmorphic features including ocular hypertelorism, upslanting palpebral fissures, telecanthus, flattened nasal bridge, micrognathia and prognathism are distinctive.[7] Cleft palate and uvula bifida were reported in some patients. Dental anomalies are also frequent. Atypical body shape including narrow shoulders and chest, inverted

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nipples and short neck are characteristic.[8] Difficult intubation in these patients is anticipated due to facial dysmorphism and malformations. For this reaseon we considered video laryngoscope for possible difficult airway management. Although we anticipated difficult intubation-ventilation in our patient due to micrognathia and narrow mouth opening, we didn't facy any problem with mask ventilation or using direct laryngoscope.

49XXXXY syndrome is mainly characterized by mental retardation, skeletal anomalies e.g radioulnar synostosis and gental anomalies e.g hypogonadism.[7] Hypotonia, delayed motor development and short stature are also seen. Hypotonia was reported in 33% of the patients.[9,10]Our patient had ambiguous genitalia, mental retardation and radioulnar synostosis at birth. He still has delays in his motor and language development.

To our knowledge, there are no published reports regarding anesthesia management of 49, XXXXY patients. Due to characeristic skeletal and muscular features in this syndrome, we applied total intravenous anesthesia using propofol-ramifentanil rather than using inhaler agents. Our patient was hemodynamically stable during the operation and no postoperative anesthetic complications were reported.

Congenital heart diseases are seen in 14% of the patients; patent ductus arteriosus (PDA) is the most common defect.[11,12] Unoperated or residual shunt lesions affect the clinical outcome of the patients. Atrial septal defect (ASD), ventricular septal defect (VSD)and patent ductus arteriosus (PDA), increased pulmonary blood flow, congestive heart failure, increased pulmonary resistance and pulmonary hypertension that may affect anesthesia management are common.[13] Our patient's ASD closed spontaneously without any surgical intervention.

Hypogonadism, infertility, deep vein thrombosis, pulmonary emboli and type 2 diabetes mellitus are detected during 49XXXXY patients follow-up. Although thromboembolic complications are usually detected in adults, children with 49XXXXY syndrome have increased risk of life threatening thromboembolic complications that should be taken into consideration during preoperative management of the patient.[14,15] Fortunately, our patient had no risk factors for thromboembolism.

In conclusion, general anesthesia is frequently required in patients with 49XXXXY syndrome before surgical interventions due to high frequency of genital system and other systems anomalies. Cardiac and facial malformations significantly impact airway management. Preoperative cardiac assessment of patients is a must to identify unstable or undiagnosed conditions. Risk of difficult mask ventilation and intubation should be anticipated in patients with dental and orthodontic anomalies and proper preparations should be made. Special attention must be paid to prevention and control of thromboembolic complications.

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