Case Report/*Olgu Sunumu*

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General Anesthesia Management of a Case with Trisomy 8 Mosaic Syndrome: A Rare Chromosomal Anomaly and Anesthesia Difficulties

Trizomi 8 Mozaik Sendromlu Bir Vakanın Genel Anestezi Yönetimi: Nadir Bir Kromozomal Anomali ve Anestezi Zorlukları

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ABSTRACT

Trisomy 8 Mosaic Syndrome (T8MS) is a rare genetic disease. Patients with congenital anomalies carry some risks in terms of anesthesia. Although the anesthesia difficulties of patients with congenital anomalies are well described in the literature, studies or case reports presenting patients with trisomy 8 are quite limited. The case presented here is a 6-year-old boy weighing 22 kg. Since the patient had some multiple abnormalities such as retromicrognathia, high palate and short neck, the operating table was prepared in accordance with the hospital's Difficult Airway Protocol on the day of surgery. The patient was intubated without any complications. It is well known anesthesia difficulties on patient with chromosomal abnormalities. Although the T8MS patient has multiple congenital anomalies, taking appropriate precautions for difficult airway can provide safe anesthesia to the patient.

Keywords: Trisomy 8, difficult airway, pediatric patients, chromosomal abnormalty, congenital anomaly, general anesthesia

ÖZ

Trizomi 8 Mozaik Sendromu (T8MS) nadir görülen bir genetik hastalıktır. Konjenital anomalili hastalar anestezi açısından bazı riskler taşımaktadır. Konjenital anomalili hastaların anestezi zorlukları literatürde iyi tanımlanmış olmasına rağmen, trizomi 8'li hastaları sunan çalışmalar veya olgu sunumları oldukça sınırlıdır. Burada sunulan vaka 22 kg ağırlığında 6 yaşında bir erkek çocuktur. Hastada retromikroginatti, yüksek damak ve kısa boyun gibi bazı çoklu anormallikleri olması nedeni ile ameliyat günü hastanenin Zor Havayolu Protokolü doğrultusunda ameliyat masası hazırlandı. Hasta komplikasyonsuz bir şekilde entübe edildi. Kromozomal anormallikleri olan hastalarda anestezi zorlukları iyi bilinmektedir. T8MS hastasının birden fazla konjenital anomalisi olmasına rağmen, zor havayolu için uygun önlemlerin alınması hastaya güvenli anestezi sağlayabilir.

Anahtar sözcükler: Trizomi 8, zor havayolu, pediatrik hastalar, kromozomal anormallik, konjenital anomali, genel anestezi

INTRODUCTION

Trisomy 8 Mosaic Syndrome (T8MS), which is also known as Warkany Syndrome, is detected with an incidence of 1 in 20.000-50.000 births as a rare genetic condition considered to occur as a result of Postzygotic Mitotic Nondisjunction (1,2). Complete Trisomy 8 cases are generally considered fatal, and therefore, Trisomy 8 cases are often believed to be mosaic (3).

The clinical manifestation of T8MS might be quite diverse. It might be accompanied by several developmental anomalies such as dysmorphic facial findings, mental retardation at different levels, corpus callosum agenesis, cardiac pathologies, deep palmar-plantar striations, hydrocephalus, and generally represents a heterogeneous clinical manifestation (4,5). A limited number of cases have been presented in the literature to date, and to our knowledge, case reports on anesthetic management of patients with T8MS are even more limited. For this reason, the presentation of this case was found to be interesting.

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CASE REPORT

A 6-year-old male patient weighing 22 kg, who was scheduled to undergo dental intervention under general anesthesia, was evaluated at the anesthesia clinic. Due to multiple tooth decay, 3 tooth extractions and 2 tooth fillings were planned. The patient had hypertelorism, flattened nasal root, deep-seated eyes, microphthalmia, dysplastic ears, full nasal tip, retromicrognathia, separated nipples, long body structure, deep palm-foot lines, limitation of first metacarpophalangeal joint movements in the hands, and clinodactyly in the fourth toes (Figure 1). No additional pathology was detected in the hemogram, coagulation test, and physical examination of the patient, whose medical history revealed T8MS. The patient was consulted preoperatively with the Pediatrics Clinic and Pediatric Cardiology Clinic, which did not make additional recommendations. No cardiac pathology was observed in the patient.

The patient was taken to the surgery room after a 6-hour fasting period and monitored with electrocardiography, non-invasive arterial pressure, and pulse oximetry on the day of the surgery. The patient's vital signs on admission were as follows: heart rate 100 bpm, blood pressure 90/50 mmHg, SpO_2 value 98%. Hemodynamic parameters were stable in the perioperative period and no clinical intervention was needed.

The surgery table was prepared in line with the hospital's Difficult Airway Protocol on the day of the surgery. After the appropriate fasting period, the patient was given 0.5 mg kg^{-1} oral midazolam half an hour before the surgery and taken to the surgery room. Standard monitoring was applied. The patient was preoxygenated with a 10 L min^{-1} face mask for 5 minutes. Anesthesia was induced with 2.5 mg kg^{-1} intravenous (iv) propofol, 1 µg kg^{-1} iv fentanyl, and 0.6 mg kg^{-1} rocuronium. The oral airway was used due to difficulty during mask ventilation. After 2 minutes, the patient underwent nasotracheal intubation with a size 4.5 spiral tube using a video laryngoscope. Anesthesia was maintained with a mixture of 50% air + 50% O_2 and the minimum alveolar concentration (MAC) value was monitored to be between 1-1.5. Fresh gas flow was applied as 4 L min^{-1}. Paracetamol 10 mg kg^{-1} was administered iv 10 min before extubation. After the 40-min surgery, the muscle relaxant was reversed with 4 mg kg^{-1} iv sugammadex, and the patient was awakened without complications. The patient, who had no respiratory or hemodynamic complications in the post-operative follow-up room, was discharged the next day.

DISCUSSION

In the anesthesia management of T8MS patients, the key points include congenital heart diseases, urinary system pathologies, presence or absence of hematological diseases, mask ventilation, and endotracheal intubation difficulties. The T8MS case we presented is unique as it is the first reported in the literature involving a pediatric patient who was nasally intubated.

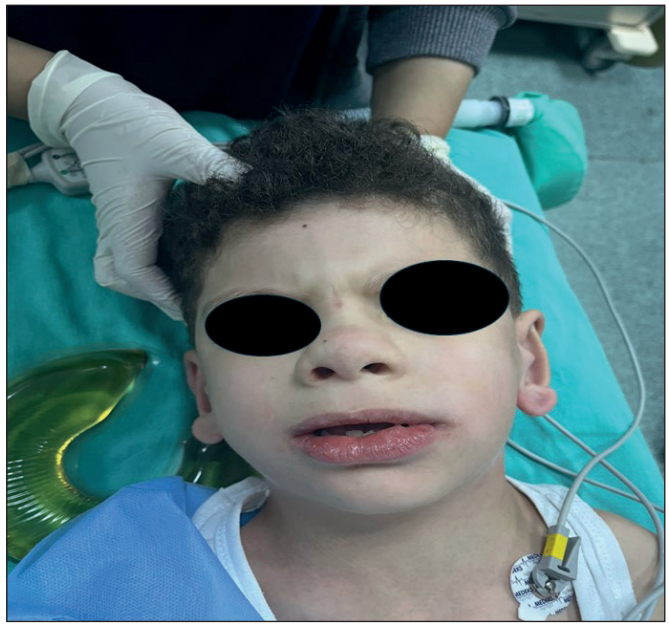

Figure 1. Microphthalmia, dysplastic ears, full nasal tip, retromicrognathia.

Craniofacial anomalies (micrognathia, bulbous nose, thick lips, etc.), long bones, and skeletal system anomalies are important risk factors for difficult intubation in T8MS cases (1, 6). In our case, bulbous nose, flattened nasal root, dysplastic ears, thick lips and prominent forehead were detected. These anatomical variations may make it difficult to perform mask ventilation and intubation on the first attempt. Tracheal structures may be traumatized after each attempt. Our patient had retromicrognathia. In this case, the oral airway was used due to difficulty during mask ventilation. Our patient's nasal-tracheal intubation was successful on a single attempt.

In patients with expected difficulty in airway control, successful intubation at one time will reduce the risk of traumatic complications. Various clinical methods have been developed to increase success. The literature suggests various alternative methods (fiberoptic laryngoscopy, video laryngoscopy, etc.) for patients with potential difficult airways (7).

Difficult airway in pediatric patients may be more complex due to children's anatomical and physiological differences. Therefore, specially developed equipments and techniques should be used for pediatric cases. Craniofacial anomalies should be considered in pediatric patients with chromosomal anomalies, such as T8MS. Thinner endotracheal tubes,

appropriately sized laryngoscope blades or advanced techniques such as videolaryngoscope or fiberoptic bronchoscopes should be used (8,9).

Very few cases are reported in the literature regarding anesthesia management of patients with T8MS. However, craniofacial anomalies that may cause difficult airway and intubation are emphasized in these cases. Kang et al. reported their experience of anesthesia management in a case diagnosed with T8MS with myelodysplastic syndrome. This study emphasized the importance of careful preoperative evaluation and compliance with difficult airway protocols (10).

Congenital cardiac pathologies such as pulmonary stenosis, heart wall defects, and persistent ductus arteriosus might also be detected in approximately 25% of cases (9). Knowing cardiac pathologies in the preoperative period is very important for choosing the appropriate anesthesia method and adequate monitoring. The cardiac examination of the patient was performed carefully and no cardiac pathology was detected.

Another important characteristic of T8MS cases is the increased risk of hematological diseases. The incidence of diseases such as Myelodysplastic Syndrome, Aplastic Anemia, Acute Myelocytic Leukemia, and Chronic Myelocytic Leukemia is higher in approximately 10% of patients compared to the normal population (5). For this reason, hematological examination must be performed with care and these patients must be monitored periodically. No pathology was detected in the hematological laboratory examinations performed in our case.

Hematological diseases, urinary system malformations and diseases, corpus callosum agenesis, mental retardation, and developmental delay might also be detected in patients with T8MS (3,5,9). Therefore, preoperative evaluation is of vital importance.

General anesthesia in pediatric patients with T8MS requires careful preparation and attention due to anatomical and physiological differences. This case emphasizes that the possibility of difficult intubation in a patient with T8MS should be considered and that the anesthesia team should be prepared for such cases. A detailed pre-anesthetic evaluation of patients with T8MS, provision of appropriate intubation equipment, and availability of alternative airway management methods when necessary are critical for successful anesthesia management. Such case reports may contribute to understanding the difficulties encountered in T8MS and similar rare syndromes and guide future clinical practice.

AUTHOR CONTRIBUTIONS

Conception or design of the work: KK, ET
Data collection: KK
Data analysis and interpretation: KK, ET
Drafting the article: KK, ET
Critical revision of the article: ET
The author (KK, ET) reviewed the results and approved the final version of the manuscript.

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