Case Report/*Olgu Sunumu*

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Perioperative Anesthesia Management in Spinal Surgery in a Patient with Autosomal Recessive NDUFV1 Gene Mutation

Otozomal Resesif NDUFV1 Gen Mutasyonu Olan bir Hastada Omurilik Cerrahisinde Perioperatif Anestezi Yönetimi

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ABSTRACT

Mitochondrial diseases are rare disorders associated with high mortality rates, particularly in childhood. Energy production in organs with high metabolic demands is primarily dependent on mitochondrial oxidative phosphorylation. Impaired oxidative phosphorylation and limited metabolic reserves lead to an increased risk of complications in this patient group during the perioperative period. Clinical findings usually appear during childhood and may include muscle weakness, lethargy, seizures, hypotonia, dysphagia, and visual impairment. Involvement of the central nervous system, cardiovascular system, skeletal muscles, and other organ systems results in increased sensitivity to anesthetic agents. Therefore, considering the depressant effects of general anesthetics, anesthesia management in individuals with mitochondrial disease is more complex and challenging. Mitochondrial enzyme deficiencies can develop due to numerous genetic and environmental factors. Ensuring normothermia, normoglycemia, and adequate respiratory support during the perioperative period are fundamental principles. However, optimal anesthesia strategies in adult patients with mitochondrial disease have not yet been clearly defined. This case presentation describes the perioperative anesthesia management of an adult patient diagnosed with Mitochondrial Complex I Deficiency, Nuclear Type 4 (c.707T>C [p.Phe236Ser], homozygous) due to an autosomal recessive NDUFV1 gene mutation. It was determined that the patient's parents are related, there is an absence of documented cases of mitochondrial disease within the family, and the patient had previously received a diagnosis of Alexander disease. Due to the progression of clinical symptoms, spinal stabilization surgery was planned. The aim of this case report is to discuss the perioperative anesthetic management and clinical challenges encountered in a rare subtype of mitochondrial enzyme deficiency for which anesthetic management has not previously been reported, in the context of the existing literature.

Keywords: Mitochondrial diseases, anesthetic drugs, adult patients

ÖZ

Mitokondriyal hastalıklar nadir görülen bozukluklar olup, özellikle çocukluk çağında yüksek mortalite oranları ile ilişkilidir. Yüksek metabolik gereksinime sahip organların enerji üretimi esas olarak mitokondriyal oksidatif fosforilasyona bağımlıdır. Oksidatif fosforilasyonun bozulması ve sınırlı metabolik rezervler, bu hasta grubunda perioperatif dönemde artmış komplikasyon riskine yol açmaktadır. Klinik bulgular genellikle çocukluk çağında ortaya çıkar ve kas güçsüzlüğü, letarji, nöbetler, hipotoni, disfaji ve görme bozukluklarını içerebilir. Merkezi sinir sistemi, kardiyovasküler sistem, iskelet kasları ve diğer organ sistemlerinin etkilenmesi, anestezik ajanlara karşı artmış duyarlılık ile sonuçlanır. Bu nedenle genel anesteziklerin baskılayıcı etkileri göz önünde bulundurulduğunda, mitokondriyal hastalığı olan bireylerde anestezi yönetimi daha karmaşık ve zorlayıcıdır. Mitokondriyal enzim eksiklikleri çok sayıda genetik ve çevresel faktöre bağlı olarak gelişebilir. Perioperatif dönemde normoterminin, normogliseminin ve yeterli solunum desteğinin sağlanması temel ilkeler arasında yer alır. Ancak erişkin mitokondriyal hastalarda optimal anestezi stratejileri henüz net olarak tanımlanmamıştır. Bu olgu sunumunda, otozomal resesif NDUFV1 gen mutasyonu sonucu Mitokondriyal Kompleks I Eksikliği, Nükleer Tip 4 (c.707T>C [p.Phe236Ser], homozigot) tanısı almış erişkin bir hastada uygulanan perioperatif anestezi yönetimi sunulmaktadır. Hastanın ebeveynlerinin akraba olduğu, aile öyküsünde mitokondriyal hastalık bulunmadığı ve daha önce Alexander hastalığı tanısı ile takip edildiği öğrenilmiştir. Klinik semptomların progresyonu nedeniyle spinal stabilizasyon cerrahisi planlanmıştır. Bu olgu sunumunun amacı, daha önce anestezi yönetimi bildirilmemiş nadir bir mitokondriyal enzim eksikliği alt tipinde perioperatif anestezi yaklaşımını ve karşılaşılan klinik zorlukları literatür eşliğinde tartışmaktır.

Anahtar sözcükler: Mitokondriyal hastalıklar, anestezik ilaçlar, erişkin hastalar

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INTRODUCTION

Mitochondrial diseases (MD) encompass disorders arising from mitochondrial dysfunction and genetic mitochondrial defects, which can affect various organ systems such as the heart, brain, skeletal muscle, sensory organs, and endocrine cells. Once considered a rare clinical condition, MDs are now known to be a significant cause of a wide variety of disorders. Surgery and anesthesia in children or adults with mitochondrial defects are becoming increasingly common (1). Reports on anesthesia-related issues in adult patients with MD are even less common than in pediatric patients, making the preferred anesthesia management strategy in these patients unclear. These diseases typically cause impaired energy metabolism, leading to metabolic acidosis and cellular damage. Since organ systems with high metabolic demands are dependent on energy supplied by mitochondria, MDs cause various physiological changes that affect the perioperative period (2,3). With this case presentation, we aim to share our experience with perioperative anesthesia management in an adult patient diagnosed with Mitochondrial Complex I Deficiency at our clinic.

CASE PRESENTATION

A 37-year-old male patient (body mass index: 24.73 kg m^{-2}) American Society of Anesthesiologists (ASA) II, presented to the neurosurgical clinic due to a marked increase in his complaints of back and bilateral hip pain over the preceding week, which had been persistent for a period of two months. The patient was scheduled for lumbar stabilization and transforaminal lumbar interbody fusion surgery.

The patient had been followed up with a diagnosis of Alexander disease due to progressive vision loss starting at age seven, spasm affecting breathing accompanied by vomiting, periodic lethargy, and apnea attacks; and the presence of periventricular symmetric cystic areas on cranial magnetic resonance imaging scans. In 2003 (at age 16), the patient underwent vertebral stabilization surgery for kyphoscoliosis, and in 2019 (at age 32), the patient required mechanical ventilation with orotracheal intubation in the intensive care unit due to apnea attacks and the subsequent development of respiratory failure. In 2023, Sanger sequencing analysis performed at the genetic disease evaluation center revealed a diagnosis of Nuclear Type 4 Mitochondrial Complex I Deficiency associated with an autosomal recessive NDUFV1 gene mutation. A homozygous c.707T>C (p.Phe236Ser) mutation was detected in exon 6 of the NDUFV1 gene (NM_007103.4), and the variant was classified as “likely pathogenic”.

During the preoperative assessment, the patient was conscious, cooperative, and oriented. The Glasgow Coma Scale score was 15. Upper extremity muscle strength was bilateral 5/5, distal lower extremity muscle strength was 5/5, and proximal muscle strength was 4/5. Transthoracic echocardiography performed to rule out possible cardiomyopathy showed a left ventricular ejection fraction of 62%.

The fasting period was 6 hours, during which the basal metabolic requirement was met intravenously with a 5% dextrose–0.45% NaCl solution at a rate of 100 mL h^{-1}. For premedication, 2.5 mg midazolam intravenous (IV) (0.04 mg kg^{-1}) was administered, and the patient was taken to the operating room with 3 L min^{-1} oxygen support via a nasal cannula. Anesthesia induction was performed with midazolam 4 mg IV, lidocaine 60 mg IV, propofol 35 mg IV (0.5 mg kg^{-1}), fentanyl 100 µg IV (1.5 µg kg^{-1}), and rocuronium 50 mg IV (0.6 mg kg^{-1}). Following induction of anesthesia, arterial cannulation was performed via the left radial artery under ultrasound guidance for intra-arterial monitoring. When the right internal jugular vein was evaluated by ultrasound prior to central venous catheterization, it was observed that the vein showed bifurcation proximally and had a narrow lumen diameter (Figure 1). Catheterization was successfully performed via the left internal jugular vein, as it had a wider diameter. The patient was placed in the prone position and active warming was provided using warm air blanket. Anesthesia maintenance was achieved with 2% sevoflurane, remifentanil infusion (0.05–0.1 µg kg^{-1} min^{-1}), and 10 mg rocuronium IV every 30 minutes. Anesthesia depth was continuously monitored using Bispectral Index with Density Spectral Array (BIS-DSA, former Covidien, now Medtronic plc, Ireland). During the operation, the basal fluid requirement was met with 5% dextrose–0.45% NaCl at a rate of 100 mL h^{-1}; additional fluid requirements were met with crystalloid solutions containing balanced electrolytes. The initial arterial blood gas analysis showed a lactate level of 1.7 mmol L^{-1}, and hourly arterial blood gas analyses were monitored throughout the intraoperative course. The lactate level was monitored throughout the operation and ranged between 0.6 and 2.0 mmol L^{-1}. At the request of the neurosurgery team, 250 mg of methylprednisolone IV was administered for anti-edema purposes, and 1 g of tranexamic acid IV was administered to reduce perioperative bleeding. Within the scope of multimodal analgesia, 1 g paracetamol IV, 50 mg dexketoprofen IV, 5 mg morphine IV, and 4 mg ondansetron IV were administered. Rocuronium was administered in a weight-based, intermittent dosing regimen, and neuromuscular blockade was reversed with sugammadex at the end of surgery. The patient was successfully extubated without complications, suggesting adequate reversal of neuromuscular blockade. The total anesthesia time was 270 minutes, and the surgical time was 205 minutes. Postoperative analgesia was intended to be provided by IV patient controlled analge-

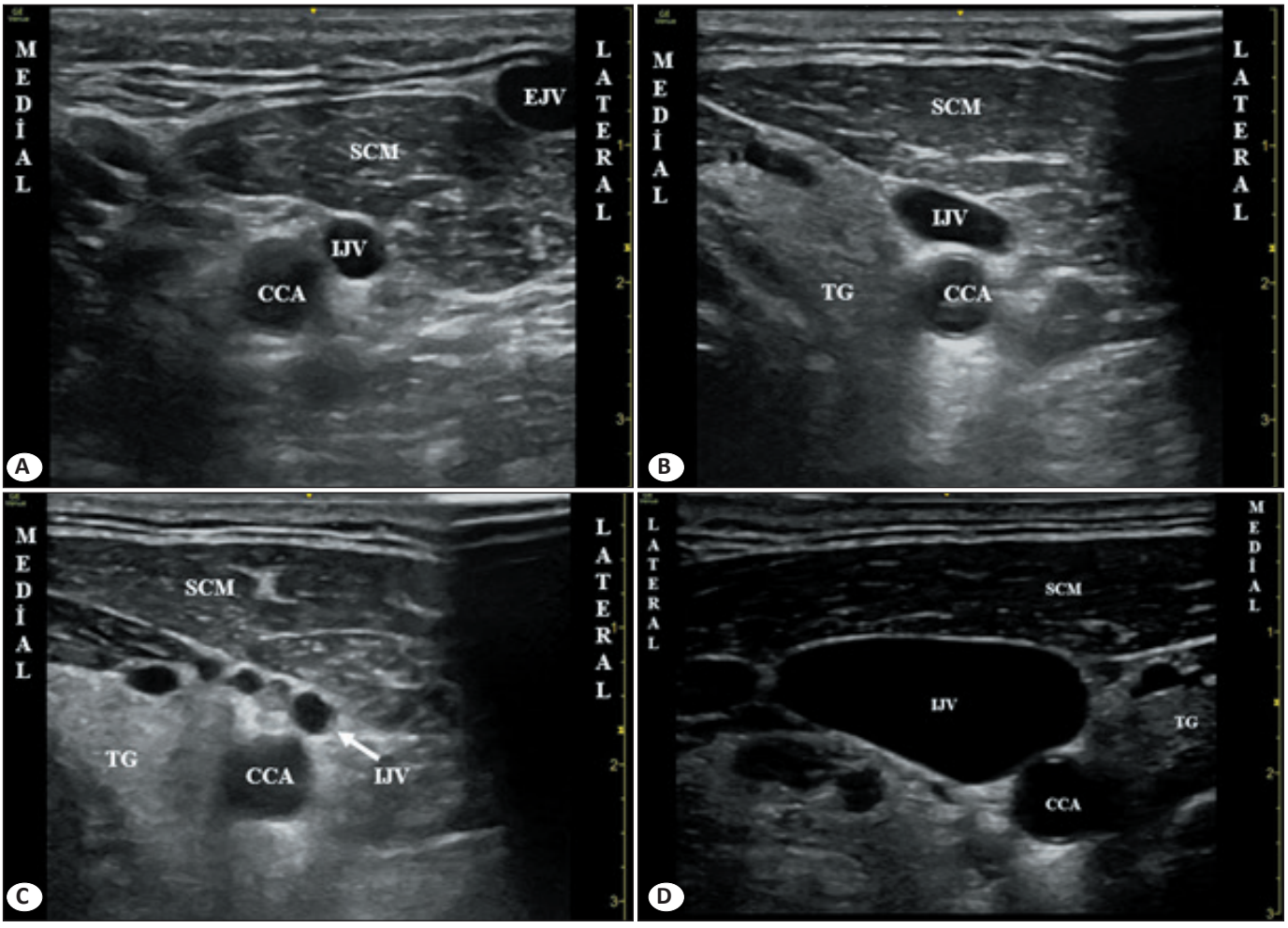


Figure 1. A-C) Imaging of the right jugular vein from proximal to distal using a linear ultrasound probe. **D)** Imaging of the left jugular vein using ultrasound.
IJV: Internal jugular vein, **SCM:** Sternocleidomastoid muscle, **TG:** Thyroid gland, **CCA:** Common carotid artery.

sia. Total intraoperative fluid replacement was calculated as 2500 mL, estimated blood loss as 300 mL, and urine output as 1000 mL. The lowest recorded body temperature was 35.6°C. To restore and maintain normothermia, multiple warming strategies were employed, including a forced-air warming blanket, warmed IV fluids, increased operating room temperature, and heated humidified airway gases, after which normothermia was successfully maintained. The patient was transferred to the intensive care unit for postoperative monitoring. Blood samples taken two hours before the operation showed creatine kinase (CK): 2546 U L^{-1}, CK-MB: 71.8 U L^{-1}, and high-sensitivity troponin T: 8.05 ng L^{-1}. Postoperative laboratory tests revealed CK: 1135 U L^{-1}, CK-MB: 37.9 U L^{-1}, high-sensitivity troponin T: 8.98 ng L^{-1}, and lactate: 2.4 mmol L^{-1}. The elevated preoperative and postoperative CK levels were consistent with the patient's underlying chronic mitochondrial myopathy. To exclude pigment nephropathy-related acute kidney injury, renal function tests (creatinine and blood urea nitrogen) were closely monitored throughout the perioperative period. Urine output was measured hourly and urine color was observed; no discoloration suggestive of myoglobinuria was detected. The patient was monitored in the intensive care unit for 24 hours with 2–3 L min^{-1} nasal cannula oxygen support. Blood glucose and lactate levels were closely monitored via arterial blood gas analyses. Normoglycemia (70–140 mg dL^{-1}) was maintained as the glycemic target throughout the perioperative period. As vital signs remained stable and respiratory support was not required, the patient was transferred to the neurosurgery ward and discharged on the 8th postoperative day with a favorable prognosis.

Written informed consent was obtained from the patient for the publication of this case report and accompanying images in accordance with the journal's ethical guidelines.

DISCUSSION

In individuals with mitochondrial disease, caution should be exercised regarding lactic acidosis during the perioperative period, preoperative organ dysfunction, increased sensitivity to muscle relaxants, and thermoregulatory disorders. Data on anesthesia management in the adult patient group are limited, and results from existing case series regarding inhalation and IV anesthetics are heterogeneous; the most frequently reported complication is lactic acidosis (1). The general approach involves the use of anesthetic agents at low doses, avoiding prolonged fasting, and preventing hypoglycemia, postoperative nausea and vomiting, and hypothermia. The relationship between mitochondrial function and the response to anesthetic agents has not yet been fully elucidated. Both total IV anesthesia and inhalation anesthesia can be safely titrated using processed electroencephalogram monitoring such as Bispectral Index (BIS). In our patient, BIS-DSA monitoring provided a real-time visual representation of raw electroencephalogram patterns, enabling continuous detection of burst suppression and allowing precise titration of anesthetic agents to avoid cortical overdosing. This was particularly valuable given that mitochondrial complex I deficiency impairs neuronal oxidative phosphorylation, potentially increasing susceptibility to anesthetic-induced cerebral metabolic depression. In this case, 5% dextrose–0.45% NaCl was preferred throughout the perioperative period to prevent hypoglycemia and meet basal metabolic requirements, in line with the recommendation for lactate-free, dextrose-containing IV fluids (2–4). Although mitochondrial cardiomyopathy can be associated with structural or functional abnormalities of the myocardium due to defects in the mitochondrial respiratory chain, our patient's electrocardiography and echocardiography findings were normal. Although it has been reported that volatile anesthetics such as isoflurane, desflurane, and sevoflurane may interact adversely with mitochondrial myopathies, the elimination of these agents via the respiratory tract and their minimal contribution to metabolic load may constitute a potential advantage over IV agents (3). Propofol is known to inhibit mitochondrial respiratory chain complexes I and IV, thereby impairing oxidative phosphorylation and potentially precipitating propofol infusion syndrome, which is characterized by metabolic acidosis, rhabdomyolysis, and cardiac failure. Given this mechanism, prolonged propofol infusion for anesthesia maintenance was deliberately avoided in our patient. Instead, propofol was administered solely at an ultra-low induction dose (35 mg, 0.5 mg kg^{-1}), and anesthesia maintenance was achieved with sevoflurane, thereby minimizing the risk of mitochondrial toxicity. Despite the presence of contradictory data in the literature regarding the selection of anesthetic agent, no evidence has been provided to suggest that any particular agent is unequivocally contraindicated (5–8). Anesthesia management administered at low doses, individualized, and closely monitored, appears to be a safe approach in such cases. In this case, it was also observed that the left radial artery and right internal jugular vein were smaller in diameter than their contralateral counterparts. Although no definitive conclusions regarding the prevalence of anatomical variations in mitochondrial diseases can be drawn from a single case, this observation reinforces the importance of routine ultrasound guidance during invasive vascular procedures to ensure patient safety in the presence of individual anatomical challenges. Ensuring patient safety during fasting by supporting metabolic requirements with dextrose- containing crystalloids, maintaining strict glucose and temperature control, and close monitoring during the postoperative period are of great importance.

CONCLUSION

This case of NDUFV1 mutation-associated mitochondrial complex I deficiency underscores the clinical importance of identifying specific genetic mutation subtypes, as precise diagnosis directly informed our individualized anesthetic strategy and contributed to a favorable outcome. Large-scale case series evaluating anesthesia approaches across different mitochondrial subtypes are needed to establish evidence-based perioperative protocols. Individualized anesthesia management, as demonstrated in this case, remains a key determinant in reducing the risk of perioperative complications.

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AUTHOR CONTRIBUTIONS

Conception or design of the work: ROG, ZG
Data collection: ROG, HE
Data analysis and interpretation: MSS
Drafting the article: HE, MSS
Critical revision of the article: ZG
Other (study supervision, fundings, materials, etc): -
The authors (ROG, ZG, MSS, HE) reviewed the results and approved the final version of the manuscript.

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