



Case Report

An Anaesthetic Approach To A Case With Emery-Dreifuss Muscular Dystrophy

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Abstract

A 13-year-old girl with known Emery-Dreifuss Muscular Dystrophy (EDMD) admitted to the emergency service and diagnosed as appendicitis. On physical examination, contractures of her elbows, Achilles tendons and posterior cervical muscles were noticed. She showed muscular wasting. Preoperative airway examination revealed a difficulty for endotracheal intubation. Following the induction with fentanyl (1 µg/kg) and propofol (3 mg/kg), an attempt for laryngoscopy was done without using a neuromuscular blocking agent. Intubation seemed to be apparently very difficult and a laryngeal mask was inserted. Anaesthesia was maintained with desflurane 4-5 % and 67 % nitrous oxide in O₂. Neuromuscular block was established with atracurium (0.5 mg/kg) after being sure that airway was secured with the laryngeal mask. The blood pressure, heart rate and oxygen saturation of the patient were stable all through the operation. The patient recovered from neuromuscular block without administration of anticholinesterase. In conclusion, this case report illustrates the potential difficulty of endotracheal intubation. Desflurane anaesthesia combined with atracurium offers an uneventful peri- and postoperative course.

Keywords: muscular dystrophy, Emery-Dreifuss, anaesthetic techniques: inhalational anaesthetics

Emery-Dreifuss Musküler Distrofili Bir Olguda Anestezik Yaklaşım

Özet

Acil servise başvuran, Emery-Dreifuss mskler distrofisi (EDMD) olduėu bilinen 13 yařındaki kız olguya apendisitis tanısı kondu. Fizik muayenede, dirseklerde, Ařil tendonu ve posteriyor servikal kaslarda kontraktr dikkati ekiyordu. Olgu kas yorgunluėu belirtileri gsteriyordu. Operasyon ncesi yapılan havayolu muayenesi, endotrakeal entbasyonun zor olacaėını iřaret ediyordu. Fentanyl (1 µg/kg) ve propofol (3 mg/kg) indksiyonu sonrası, kas gevřetici ajan kullanılmadan laringoskopi denendi. Entbasyonun belirgin zor olduėu izlendi ve laringeal maske yerleřtirildi. Anesteziye, oksijen iinde 4-5 % desflurane ve 67 % nitroz oksit ile devam edildi. Laringeal maske ile havayolu gvenceye alındıktan sonra, atracurium (0.5 mg/kg) ile nromskler blok oluřturuldu. Operasyon sresince olgunun kan basıncı, kalp hızı ve oksijen saturasyonu normal sınırlarda seyretti. Hastada oluřturulan sinir-kas bloėu, antikolinesteraz verilmeden ortadan kalktı. Desflurane ve atracurium anestezisinin herhangi bir yan etkiye neden olmadıėı izlendi. Sonu olarak, bu olgu sunumu olası endotrakeal entbasyon glėn ortaya koymaktadır. Atrakuryum ile birlikte desfluran anestezisi olaysız bir peri- ve postoperatif seyir saėlamaktadır.

Anahtar Kelimeler: mskler distrofisi, Emery-Dreifuss, anestetik teknikler: inhalasyon anestetikleri

Introduction

Muscular Dystrophies are a group of primary muscular atrophies characterized by degeneration of the muscle fibres without any evidence of denervation. Emery-Dreifuss Muscular Dystrophy (EDMD) is the third most common of the X-linked recessive muscular dystrophies. The mutations of the STA gene at Xq28, which encodes the nuclear membrane protein emerin cause this dystrophy by disappearance of emerin completely in muscle^{2,3}. However, there have been some well documented sporadic cases in girls too^{9,12,14}. The autosomal dominant form of Emery-Dreifuss muscular dystrophy is caused by mutations in the gene encoding for the lamins A and C. These cases were labelled Emery-Dreifuss muscular dystrophy phenotype¹⁴. We present a case with EDMD who has been planned for appendicectomy under general anaesthesia with desflurane.

Case Presentation

A 13-year-old girl (36 kg, 142 cm) with known EDMD admitted to the emergency service of our hospital with the complaint of abdominal pain. She has been diagnosed as having EDMD at the Department of Neurology, University of Istanbul, Capa, Turkey. After being observed for appropriate duration by the paediatric surgeon, she got the diagnosis of acute appendicitis, and presented for appendicectomy.

On physical examination, contractures of elbows, Achilles tendons and posterior cervical muscles were noticed. There were contractures of the lower back, making her forward flexion impossible. She showed muscular wasting. She was toe walking. Beyond these, the only positive finding was direct/indirect Rebound test. Her airway was Mallampati Class IV (Fig. 1),

with a markedly reduced neck extension (Fig. 2). She had nothing significant in her family history with special interest to adverse drug reactions or anaesthetic complications. Her blood pressure was 120/80 mmHg, and pulse rate was regular at 110/min. The results of the laboratory tests were all in normal ranges except WBC: $10.7 \times 10^3/\mu\text{L}$. The creatine kinase (CK) level of the patient was 104 U/L (normal ranges: 26-174 U/L) and remained in normal ranges after the operation. The cardiological consultation of the patient held preoperatively, included echocardiographycal examination, for the possible necessity of pacemaker implantation. The results of the cardiac evaluation did not present any atrial conduction defects, ventricular involvement or cardiac failure.

Without premedication, the case was taken into the operation room for routine anaesthesia monitoring of ECG, non-invasive arterial blood pressure measurements and, pulse oxymetry. End expiratory CO₂ pressure and concentration of volatile anaesthetics were recorded continuously (Agilent, Anesthetic gas module, M1026A, USA). The intravenous infusion of 1/3 isomix with KCl- (1 meq/kg/day) was started. Following preoxygenation with eight deep breaths of 100 % oxygen and, the induction with fentanyl (1 µg/kg) and propofol (3 mg/kg), an attempt for laryngoscopy was done without using a neuromuscular blocking agent. Intubation seemed to be apparently very difficult. A further effort for the intubation of the patient was determined as time consuming and dangerous in emergency conditions, so a laryngeal mask (no=2.5) was inserted successfully at the first attempt. She had a nasogastric tube inserted in ward during observation. Before anaesthesia induction, an attempt for gastric



Figure 1: Difficult airway management with Mallampati Classification IV.



Figure 2: Marked restriction of the head movement

content aspiration was made and an empty stomach was verified. The tube was left to be a guide during laryngoscopy, but taken out before the laryngeal mask was inserted. Anaesthesia was maintained with desflurane 4-5 % and 67 % N₂O in O₂ (Ohmeda Excel 210 SE, Madison WI, USA). More narcotic analgesics were avoided during the rest of the procedure. Tidal volume was adjusted to 10 mL/kg and respiratory rate to 14 bpm for maintaining normocapnia. Atracurium (0.5 mg/kg) was administered after being sure that airway was secured with the laryngeal mask. Neuromuscular block was monitored at the thumb using a TOF-WATCH nerve stimulator (TOF-Watch SX, Organon Teknika B.V., The Netherlands). The duration of anaesthesia was 56 minutes. The blood pressure, heart rate and oxygen saturation of the case were stable all through the operation. The neuromuscular block was resolved spontaneously in normal period. The anaesthesia was completed without any complication at the end of the operation.

Discussion

The classical triad of EDMD are contractures of the elbows, Achilles tendons and spine; wasting and weakness

with a humeroperoneal distribution and cardiac conducting defects or cardiomyopathy⁹. The presented case's physical condition was in accordance with EDMD, except for cardiac symptoms. In some forms of EDMD, the patient may show only cardiac symptoms and muscle wasting with an absence of early contractures. Atrial conduction defects and bradycardia are considered hallmark of EDMD cardiomyopathy, but they are noticed generally in the second or third decade of life⁹. This may explain the lack of cardiac symptoms in our case.

The most important features of an EDMD patient for an anaesthetist are intubation difficulty due to neck stiffness and lumbar paravertebral muscle contractures which may also cause a problem in administration of regional anaesthesia⁶. So a laryngeal mask insertion may be suitable alternative for difficult intubation. Difficult intubation was predicted from preoperative airway assessment of this patient. For this reason, an endotracheal tube with a stylet, an oropharyngeal and nasopharyngeal airway, a laryngeal mask and an emergency coniotomy set was available at the time. An experienced surgical team were also in the operation room with available

unwrapped equipment for a potential emergency tracheotomy. The use of a lightwand or a fiberoptic bronchoscope could be another alternative, which the equipment was not available at that time. Preoxygenation and avoiding any neuromuscular blocking agents in induction were the other preventions.

The patient had presented with signs and symptoms of an acute abdomen, therefore she was observed in paediatric surgery unit with a nasogastric tube. The absence of intragastric content showed us that the starvation period of the patient was enough for ventilation via a laryngeal mask. It is not clear if a delay in gastric emptying takes place in EDMD patients, but a six hour preoperative fasting and treatment with a prokinetic agent may reduce the risk of pulmonary aspiration in elective EDMD cases.

Respiratory muscle weakness may interfere with an effecting coughing mechanism and leads to retention of secretions in patients with muscular dystrophy. Degeneration of the respiratory muscles combined with kyphoscoliosis may also produce a severe restrictive pulmonary defect. Patients with decreased vital capacity appear to be at greatest risk for postoperative respiratory failure and often require mechanical ventilation. On the base of clinical examination regarding dyspnea and activity level, we decided on that the patient had an adequate pulmonary reserve preoperatively. At the end of the operation, residual neuromuscular block reassessed by a nerve stimulator to make sure that patient was free of muscle relaxant.

Although there is no certain evidence about a linkage between malignant hyperthermia and X-linked muscular dystrophies⁶, it is better not to use well-known triggering anaesthetics like halothane and succinylcholine¹⁰. For this reason, a depolarizing neuromuscular

blocking agent was not preferred in this case, although an intubation difficulty was expected. Desflurane was chosen for early recovery. This case was the second, reported in English literature, indicating normal response to atracurium after a facioscapulohumeral muscular dystrophy³. Increased sensitivity to other non-depolarizing neuromuscular blocking drugs had been common in neuromuscular dystrophies¹¹. To our best knowledge, five EDMD cases were reported previously in PubMed, having uneventful operations under spinal anaesthesia, epidural anaesthesia, enflurane anaesthesia and propofol for cardioversion or target controlled infusion^{1,6,10,12,13}.

Case reports demonstrating malignant hyperthermia in humans anaesthetized with desflurane are rare and in most of them, other trigger agents, such as succinylcholine, have also been used^{5,8}. In experimental studies, desflurane was suggested as a weak trigger of malignant hyperthermia with only slight Ca^{+2} release in skeletal muscle fibres⁷. There is a long interval (90-180 min) from exposure to the occurrence of malignant hyperthermia symptoms with desflurane^{5,7,8}.

To our opinion, the important point is that this case is the first EDMD case operated under desflurane anaesthesia without any complication. However, the short duration of anaesthesia (56 min) should be taken into account. For this reason, total intravenous anaesthesia with propofol and opioids or regional anaesthesia may be considered other proper methods for longer procedures in EDMD patients.

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