

Anesthetic Management of Lesch-Nyhan Syndrome

Case Report

Lesch-Nyhan Sendromunda Anestezik Yaklaşım Olgu Sunumu

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ABSTRACT

Lesch-Nyhan disease is a rare X-linked recessive genetic disorder caused by a deficiency of hypoxanthine-guanine phosphoribosyltransferase enzyme. The patients with Lesch-Nyhan Syndrome have several problems such as positioning and difficult intravenous cannulation because of spasticity. Also these patients have risk for bradycardia, pulmonary aspiration, convulsion and sudden death. Anesthetic implications of the patients with Lesch-Nyhan Syndrome are not well described. Here we report our anesthesia experience in a 4 years-old child with Lesch-Nyhan syndrome, who underwent cataract surgery. Patients with Lesch-Nyhan syndrome should be carefully evaluated in the preoperative period especially to determine the difficulties in venous access and positioning. These patients should be closely monitored for the potential of pulmonary aspiration, and sudden death postoperatively.

Keywords: Lesch-Nyhan syndrome, anesthesia

ÖZET

Lesch-Nyhan sendromu ender rastlanan, x-resesif, hipoksantin fosforibozil-transferaz enzim eksikliği ile karakterize, pürin metabolizması bozukluğudur. Lesch-Nyhan sendromlu hastalarda pozisyon vermede, spastisite nedeniyle intravenöz kanulasyonda zorluklar mevcuttur. Ayrıca bu hastalar bradikardi, pulmoner aspirasyon, konvulziyon ve ani ölüm riskleri de mevcuttur. Lesch-Nyhan sendromunda anestezi yaklaşımı iyi tarif edilmemistir. Bu sunuda; katarakt nedeniyle operasyon geçiren Lesch-Nyhan sendromlu 4 yaşındaki hastaya anestezi yaklaşımı sunulmuştur. Lesch-Nyhan sendromlu hastalar özellikle zor venöz kanulasyon ve pozisyon nedeniyle

preoperatif donemde dikkatlice degerlendirilmelidir. Bu hastalarin, postoperatif donemde pulmoner aspirasyon ve ani olum riskleri nedeniyle yakin takipleri gerekir.

Anahtar Kelimeler: Lesch-Nyhan sendromu, Anestezi

INTRODUCTION

Lesch-Nyhan syndrome is a rare, sex-linked recessive anomaly consisting of a deficiency in the production of hypoxanthine phosphoribosyltransferase that leads to the overproduction of purine and the accumulation of uric acid. Certain characteristic neurological features includes choreoathetosis, spasticity and mental retardation resulting in self-mutilation through biting and scratching (1). The findings of MRI revealed reduced basal ganglia volume which is consistent with the dystonic movement disorder (2). No medical treatment exists to alleviate the symptoms. Therefore, preventive measures to stop self-mutilation such as direct dental intervention are the only way to reduce the harm.

The patients with Lesch-Nyhan Syndrome have several problems like positioning in operating room and difficult intravenous cannulation because of the spasticity. Also, these patients are at risk for bradycardia, pulmonary aspiration, convulsion and sudden death. Anesthetic implications of the patient with Lesch-Nyhan Syndrome are not well described. There are only a few case reports regarding the anesthetic experience with these patients (3). This report will present our anesthetic experience in a patient with Lesch-Nyhan Syndrome who underwent cataract surgery.

CASE REPORT

A 4 years old, 15 kg weight girl was scheduled for cataract surgery under general anesthesia. Preoperative airway evaluation of the child revealed micrognathia, and a short thyro-mental

distance (Fig. 1). These findings predicted a difficult airway management. Therefore, before the anesthesia induction all equipments were organized for difficult airway management. EMLA cream has been applied onto the skin of the possible venous catheterization site. Before the anesthesia induction heart rate, blood pressure, oxygen saturation with pulse oxymeter and end-tidal carbondioxide were monitored. Following pre-oxygenation, intravenous cannulae was introduced.



Figure 1: Severe spasticity in extremities.

The patient was premedicated with 0.015 mg atropine sulfat intravenously. Anesthesia was induced with 2,5 mg/kg intravenous propofol while the patient's spontaneous respiratory efforts was assisted by a face mask. After assuring the adequate ventilation by mask, the patient was given intravenous vecuronium (0.1 mg/kg) to facilitate tracheal intubation. Laryngoscopy score has been determined to be Cormack-Lehane Class II using a direct laryngoscope. After application of cricoid pressure, endotracheal tube with stylet was introduced. Anesthesia was maintained with 60 to 160 mcg/kg/min propofol infusion, and %70 N₂O-%30 O₂. Despite padding, it was difficult to secure the extremities, and the surgical team was warned so as not to compress the extremities (Fig.2). During the surgery vital signs were stable. At the end of the operation lasted 50 minutes, residual neuromuscular block was reversed by using neostigmine and atropine, and

patient was extubated when the patient opened her eyes, and resumed adequate respiration. The extremities were safe, and no anesthetic and surgical complication were shown. Patient was transferred to the service, when she was fully awaked.



Figure 2: Patient was evaluated for difficult airway.

DISCUSSION

Lesch-Nyhan syndrome is characterized by cognitive impairment, hypotonia at rest, choreoathetosis, hyperuricaemia and the hallmark symptom of severe and involuntary self-mutilation. Specifically, sudden unexplained death, abnormalities in respiration, apnea, severe bradycardia, and an increased incidence of vomiting and chronic pulmonary aspiration may preclude this patient population from receiving anesthesia (2,4). During the anesthesia induction and maintenance, propofol was used for this case. The antiemetic properties of propofol may have decreased the incidence of postoperative nausea and vomiting and subsequent potential pulmonary aspiration of gastric contents that may be prevalent with this specific patient population (3, 5). The nausea, vomiting, or pulmonary aspiration of gastric contents were not observed in this patient.

In all patients with Lesch-Nyhan syndrome, patient positioning and intravenous access were difficult due to involuntary movements and spasticity. Associated gastrointestinal disorders,

renal impairment, seizure disorders, macrocytic anemia, and malnourishment were also very often. Perioperative proper positioning, use of protective padding, and avoidance of contact with hard surfaces are the protective precautions to avoid direct pressure on susceptible skin and peripheral nerves (6). Careful positioning of the patient with proper supporting can reduce, but may not be eliminate injuries to peripheral nerves (7). Anesthesiologist should be familiar with the anatomical traces of the peripheral nerves to protect the pressure injury during the surgical procedure.

Preoperative examination of the present case revealed a difficult airway with micrognathia, short thyro-mental distance. For that reason, spontaneous ventilation was maintained during the mask ventilation, and the muscle relaxant was applied after assuring the adequacy of the assisted ventilation.

Lesch-Nyhan syndrome is a rare disorder. We have reported our experience with one patient requiring general anesthesia for cataract surgery. Caution should be exercised in extrapolating the result of our one patient based experience as this result may not be applied to all the patients having the Lesch-Nyhan syndrome. Different surgical procedures may elicit different outcomes in those patients. A detailed preoperative evaluation should include the assessment of the airway, intravenous access, and the body position. Positioning during the operation should be planned preoperatively considering the body and extremity posture. More experience to confirm the safety of general anesthesia in these patients undergoing different surgical procedures.

In conclusion, pre-operative evaluation of a patient with Lesch-Nyhan syndrome should include a careful examination of the airway, body posture and veins to be cannulated to prevent the potential difficulties of airway management, intravenous access and positioning. The patients should be closely monitored for

the potential of pulmonary aspiration, and sudden death postoperatively. A careful preoperative evaluation and perioperative management provided a successful outcome in this case.

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