Iran J Pediatr. 2016 October; 26(5):e5479. doi: 10.5812/ijp.5479.

Published online 2016 July 31. **Case Report**

Anesthetic Management in a Child With Niemann-Pick Disease

Ebrahim Espahbodi,[1] Amir Abbas Yaghooti,[1] Abbas Ostadalipour,[1] and Shaqayeq Marashi[1,*]

[1]Anesthesiology and Critical Care, Tehran University of Medical Sciences, Tehran, IR Iran

[*]*Corresponding author*: Shaqayeq Marashi, Anesthesiology and Critical Care, Tehran University of Medical Sciences, Tehran, IR Iran. Tel: +98-9123257790, E-mail: shm1377@yahoo.com

Received 2016 January 25; **Revised** 2016 March 05; **Accepted** 2016 March 14.

Abstract

Niemann-Pick is a lipid storage disease that results from a lysosomal enzyme deficiency (sphingomyelinase). It has different presentations, and it may affect various organs such as the central nervous system, kidney, liver, and spleen. Due to the complexity of the disease, careful perianesthetic management is necessary in order to reduce the risks and sequels. As there is little evidence available in the literature regarding the anesthetic implications of such patients, in this case report we describe the anesthetic management of a two-year-old female with Niemann-Pick disease.

Keywords: Niemann-Pick, Anesthesia, Child

1. Introduction

Niemann-Pick is an autosomal recessive lysosomal storage disease that is classified into four subgroups, namely A, B, C, and D (1, 2). Types A and B involve a sphingomyelinase deficiency, while Niemann-Pick type C is associated with an alteration in the intracellular transport of cholesterol and, subsequently, its accumulation in the lysosomes (1). The features of the disease include prolonged neonatal jaundice (3), massive hepatosplenomegaly, as well as pulmonary and cardiac involvement with or without neurologic manifestations (4). Niemann-Pick type A, which is also known as the infantile form, is the most common type, and its characteristic features include hepatomegaly, brain damage, and walking, speaking, and learning problems (2). It usually results in early death because of the extensive neurological involvement (2, 4). Niemann-Pick type B is associated with organomegaly, but without severe neurological symptoms (4). In Niemann-Pick type C, there is a broad spectrum of clinical manifestations, including liver and spleen involvement and neurological problems such as seizure, progressive dementia, and dystonia. Respiratory problems have also been noted (1, 5).

Due to the complex clinical progression and multiorgan involvement, the anesthetic management of Niemann-Pick patients is challenging. However, only a few articles are available concerning the anesthetic implications of such patients (1, 5, 6). Hence, in this case report we describe the anesthetic management of a Niemann-Pick patient.

2. Case Presentation

This case report concerns a two-year-old female (weighing 8 kg) known to have Niemann-Pick disease who was scheduled for an elective closed reduction of a femoral shaft fracture under general anesthesia. The first manifestation of her condition was hepatosplenomegaly at five months of age, which is when the diagnosis was made based on blood and bone marrow findings. She also exhibited developmental delay in terms of both speaking and walking. Her past surgical history was negative. However, her past medical history was positive for repeat hospital admissions due to respiratory system infection. In terms of the preoperative assessment, abdominal sonography had shown hepatosplenomegaly, while echocardiography had demonstrated satisfactory left ventricular function. The preoperative lab data were within normal limits, except for Hbg 9.7 g/dL, PLT 125000/μL, Alk-P'se 254 IU/L, urea 13 mg/dL, Cr 3 mg/dL, and CRP 15 mg/L. In the operating room, the patient was awake and conscious. Mild suprasternal retraction was observed during the respiration (respiratory rate 28 per minute). During the lung auscultation, a bilateral crackle was heard at the base of both lungs (probably due to previous infections). The sclera was also icteric. Additionally, the abdomen was distended and mild tenderness existed during palpation over the right upper quadrant. Packed red blood cells (RBC) and platelets (PLT) were reserved for the patient the day before surgery. In the operating room, standard monitoring consisting of pulse oximetry, electrocardiography (ECG), and noninvasive blood pressure (NIBP) was instituted. The baseline

O2 saturation in the room's air was 87%, and the NIBP was 78/51 mmHg.

After preoxygenation with O2 (4 - 5 L/min) via a face mask, 16 μg fentanyl and 1 mg midazolam were injected intravenously, then the induction of anesthesia was performed using the sevoflurane inhalation technique. When the proper depth of anesthesia was achieved, a 1.5 laryngeal mask airway (LMA) was inserted while the patient was spontaneously breathing. Anesthesia was maintained using sevoflurane (3% - 4%) and remifentanil (0.5 μ/kg/min). Her vital signs were stable during the operation, which lasted about 30 minutes. At the end of the procedure, an acetaminophen suppository (125 mg) was administered for postoperative pain management and sevoflurane was discontinued. The LMA was removed after laryngeal reflexes or swallowing had returned. The patient was then transferred to the postoperative care unit (PACU) and, after fulfilling the discharge criteria, she was sent to the ward while hemodynamically stable.

3. Discussion

In general, Niemann-Pick disease can affect multiple organs, including the liver, spleen, lung, and heart, with or without neurologic deficits (4). All these features can prove challenging during the anesthetic management of such patients. Due to both the liver involvement and the probability of an alteration in drug metabolism, anesthetic drugs should be used with caution in Niemann-Pick patients. We therefore chose to use sevoflurane, which can be better tolerated in this specific condition. Additionally, its rapid washing out after discontinuation helped us to achieve rapid recovery with minimal postoperative sequel on respiration and ventilation. Similar to the approach of Araujo et al. (5), we used remifentanil as the analgesic because of its short duration of action, which provided us with a rapid and predictable recovery. Moreover, its metabolism, which is independent of liver function, rendered it a good analgesic choice for our patient. Another consideration is the possibility of thrombocytopenia due to splenomegaly. Miao et al observed thrombocytopenia in 50% of their patients (1). Our patient also had mild thrombocytopenia, so we cautiously reserved packed RBC and PLT for her prior to the operation.

Renal dysfunction was another challenge in our patient. In 2009, renal involvement in Niemann-Pick disease was reported for the first time by Grafft et al. (4). They described how renal involvement should be considered in all patients with this disease, especially those with renal dysfunction. It should be noted that none of our chosen medications negatively impact renal function. In Niemann-Pick type A, it is better to avoid the use of a muscle relaxant due to the neurologic involvement (6), so we decided to insert the LMA under deep inhalational anesthesia without muscle paralysis.

In conclusion, the wide spectrum of manifestations and the complexity of multi-organ involvement necessitate a thorough understanding of the disease in order to identify the best method of anesthesia and, hence, to reduce perianesthetic morbidity and mortality in Niemann-Pick patients.

Footnote

Authors' Contribution: Amir Abbas Yaghooti and Abbas Ostadalipour collected the data; Ebrahim Espahbodi wrote the first draft of the manuscript, and Shaqayeq Marashi revised the manuscript.

References

1. Miao N, Lu X, O'Grady NP, Yanjanin N, Porter FD, Quezado ZM. Niemann-pick disease type C: implications for sedation and anesthesia for diagnostic procedures. *J Child Neurol.* 2012;**27**(12):1541–6. doi: 10.1177/0883073812437243. [PubMed: 22378675].
2. Galehdari H, Tangestani R, Ghasemian S. New Single Nucleotide Deletion In the SMPD1 Gene Causes Niemann Pick Disease Type A in a Child from Southwest Iran: A Case Report. *Iran J Pediatr.* 2013;**23**(2):233–6. [PubMed: 23724191].
3. Imrie J, Dasgupta S, Besley GT, Harris C, Heptinstall L, Knight S, et al. The natural history of Niemann-Pick disease type C in the UK. *J Inherit Metab Dis.* 2007;**30**(1):51–9. doi: 10.1007/s10545-006-0384-7. [PubMed: 17160617].
4. Grafft CA, Fervenza FC, Semret MH, Orloff S, Sethi S. Renal involvement in Neimann-Pick Disease. *NDT Plus.* 2009;**2**(6):448–51. doi: 10.1093/ndtplus/sfp101. [PubMed: 25949377].
5. Araujo AM, Orfao JM, Machado H. Ambulatory Anesthesia In a Patient with Niemann -Pick Disease Type C. *J Anesth Clin Res.* 2015;**6**:509.
6. Dalal PG, Coleman M, Horst M, Rocourt D, Ladda RL, Janicki PK. Case Report: Genetic analysis and anesthetic management of a child with Niemann-Pick disease Type A. *F1000Res.* 2015;**4**:1423. doi: 10.12688/f1000research.7470.1. [PubMed: 26913189].