acta medica

CASE REPORT

Anesthetic management of a patient with Nager Syndrome

Filiz Banu ETHEMOĞLU[1], [MD]
Başak AKÇA[1]*, [MD]
Aysun ANKAY YILBAŞ[1], [MD]
Filiz ÜZÜMCÜGİL[1], [MD]
Özgür CANBAY[2], [MD]

1 Hacettepe University Faculty of Medicine, Department of Anesthesiology, 1 Specialist, 2 Professor

* *Corresponding Author: Başak AKÇA, MD, Hacettepe University Faculty of Medicine Department of Anesthesiology, Sihhiye, Ankara, 06100, Turkey*

e-mail: basak_kantar@yahoo.com

Received 3 August 2015; accepted 24 December 2015; published online 24 December 2015

ABSTRACT

Nager acrofacial dysostosis is an oromandibular hypogenesis syndrome characterized with limb abnormalities. Even though its phenotypic characteristics are similar to those of Treacher-Collins Syndrome, it is considered as a separate disorder. The main concern for an anesthesiologist is the facial and mandibular malformations complicating airway management. These problems may also be associated with airway obstruction in the postoperative period. Herein we present the anesthetic management of a 4-year-old male patient with Nager syndrome.

Key words: Nager Syndrome, Airway management, Cleft Palate

Introduction

Nager acrofacial dysostosis is an oromandibular hypogenesis syndrome characterized with limb abnormalities. Even though its phenotypic characteristics are similar to those of Treacher-Collins Syndrome, it is considered as a separate disorder [1-2]. The facial and mandibular features of Nager syndrome include downward slanting palpebral fissures, malar hypoplasia, high nasal bridge, cleft palate and micrognathia, and atretic external auditory canals [2]. Preaxial limb malformations include hypoplastic or absent thumbs, radial hypoplasia and short humerus [3]. The main concern for an anesthesiologist is the facial and mandibular malformations complicating airway management. These problems may be associated with airway obstruction in the postoperative period. Associated deficiencies are vertebral malformations, congenital heart defects, and preaxial or radial upper limb defects. Due to presence of facial and limb abnormalities, multiple surgeries may be required in children with Nager syndrome.

Case Presentation

A 4-year-old male patient with Nager syndrome was scheduled for cleft palate repair and bilateral tube insertion. He had hemifacial microsomia, cleft palate, tubular nose, low-set ears that were rotated backward, long philtrum, thin upper lip, micrognathia; and radial agenesis in left upper limb, aplasia in the first and fifth fingers of the left hand, ulnar hypoplasia, as well as partial syndactylia in the third and forth fingers of right hand. Airway examination revealed the presence of Mallampati IV, opening of the mouth at least two finger wide (Figure 1), a very short neck but cervical motion was normal so that we did not examine the thyromental distance. Mental retardation, hearing loss or cardiac abnormality was not present in the patient. Septum was deviated to right in the otolaryngology examination. Bilateral external auditory canals

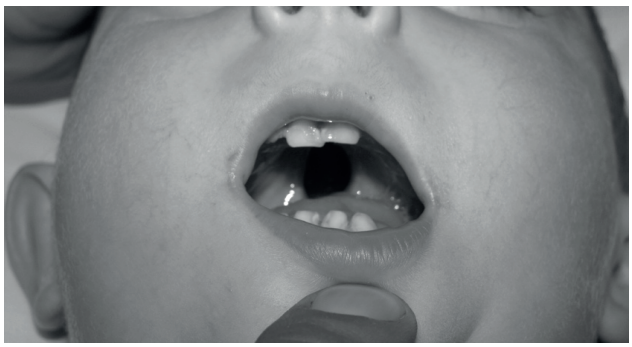

Figure 1. Four-year-old male patient with Nager syndrome.

© 2015 Acta Medica. All rights reserved.

were normal; glottic opening was intact; arytenoids were minimally prolapsed. In the preoperative period, preparations were completed for tracheotomy. Echocardiography results were normal and consistent with electrocardiogram (ECG) results. The patient had a history of delayed surgery due to difficulty of intubation, three unsuccessful intubation attempts, and arrest in the course of intubation.

The patient was premedicated with 0.5 mg/kg midazolam after 6 hours of fasting. Afterwards, he was taken to the operating theatre and was monitored in the standardized way (ECG, pulse-oximetry, noninvasive blood pressure). Ear-nose-throat (ENT) specialists were also present in the operation room in case of an airway emergency and potential need for tracheotomy during anesthesia induction. Following anesthesia induction with 8% sevoflurane, vascular access was established in the right foot with a 24 Gauge needle. For the safety of the airway, the patient was ventilated with 100% oxygen using the Rusch Nasal Airway mask. The mask was comfortable, and the patient did not have any problem regarding ventilation. After induction, sevoflurane level was decreased to 2%, thus the patient was given 3 mg/kg propofol and 0.6 mg/kg rocuronium. Prior to laryngoscopy, 0.1 mg atropine was administered to reduce secretions while 1 mg/kg of methylprednisolone was administered to prevent potential edema. The patient's trachea was intubated with spiral-embedded uncuffed 3.5 mm tube through fiberoptic bronchoscopy. Anesthesia was maintained with combination of 50% oxygen, 50% air, 2% sevoflurane together with the infusion of 0.1 mcg/kg/min remifentanil. The operation lasted 135 minutes.

No problems occurred during the operation regarding the anesthesia management, and the vital signs of the patient were stable. When the operation was completed, the patient was given 15 mg/kg paracetamol for provision of analgesia.

After the operation, the patient, whose trachea was still intubated, was transferred to the post-anesthesia care unit (PACU) for close monitoring. As the patient's general condition was good and vital signs were stable, the patient's trachea was extubated in the postoperative 2nd hour.

Discussion

Nager acrofacial dysostosis that is characterized with proximal limb dysostoses is a rare disease with less than 100 reported cases in medical literature. Characteristic facial features may be similar with the signs of the Treacher-Collins syndrome [1-2]. However, maxillary and zygomatic hypoplasia, downward slanting palpebral fissure and lower lid coloboma are more severe in the patient with Treacher-Collins syndrome. Although the aforementioned features are also seen in patients with Nager syndrome, their severity is less when compared to Treacher-Collins syndrome. Characteristic limb abnormalities (thumb/radial anomalies) of Nager syndrome are excessive mandibular hypoplasia and high incidence of lip and palate abnormalities.

Autosomal dominant, autosomal recessive, and sporadic types of the disease have been reported [4]. The recurrence risk within families is 10% in affected children and 63% in women. Also, specific chromosome abnormalities have been identified to be associated with the disease [5].

Mandibular hypoplasia, microsomiaand other organ system defects are the characteristics that may affect preoperative anesthesia management in such patients. The primary trouble for the anesthetists is the difficulty of airway management as well asmicrosomia, limitation of jaw movements, micrognathia and postoperative airway obstruction developing secondary to mandibular hypoplasia. Also, some facial deformities, such as cleft palate, cleft lip and maxillary hypoplasia, complicate airway management. Therefore, the use of an appropriate mask is crucial during mask ventilation. Similar problems have been recorded in cases with genetic disorders containing facial and oromandibular components [6]. Vener and Lerman stated the importance of airway provision in previously known genetic syndromes [7].

According to preoperative evaluation of our patient, the opening of the mouth was sufficient but he also had significant micrognathia. He had history of cancelled surgery due to difficulty of intubation, three unsuccessful intubation attempts, and arrest in the course of intubation. In literature, deaths have been reported in cases with Nager syndrome due to respiratory problems, urgent need for tracheotomy, and airway obstruction [3]. Considering the difficulty of airway provision, preoperative evaluation was conducted in detail. Tracheotomy preparation was completed for the patient, and inhalation agents were deployed for induction of the patient. Spontaneous breathing was not suppressed until the comfort of the ventilation mask was ensured. Rusch Nasal Airway was preferred for airway patency. Fiberoptic laryngoscopy was performed,

© 2015 Acta Medica. All rights reserved.

where laryngeal mask airway and tube exchangers were kept available and also we chose to use uncuffed endotracheal tube not to cause airway edema. We placed a throat pack –a bunch of sponge- covering nasopharynx to secure the airway against aspiration of the secretions. Additionally, an otolaryngology team was present at the operating theater due to the potential need for tracheotomy.

In conclusion, detailed preoperative evaluation, preparation of materials and equipments required for difficult airway, and postoperative intensive care support are important factors in ensuring successful airway management in patients with Nager syndrome.

REFERENCES

[1] **Zhang Y, Dai Y, Liu Y, Ren J.** Mandibulofacial dysostosis, microtia, and limb anomalies in a newborn: a new form of acrofacial dysostosis syndrome? Clin Genet 2010; 78: 570-4.

[2] **Jones KL.** Smith's Recognizable Patterns of Human Malformation. Philadelphia: WB Saunders, 1997: 258–9.

[3] **Danzinger I, Brodsky L, Perry R,Nusbaum S, Bernat J, Robinson L.** Nager's acrofacial dysostosis: case report and review of the literature. Int J Pediatr Otorhinolaryngol 1990; 20: 225–40.

[4] **Hecht J, Immken L, Harris L, Malini S, Scott Cl Jr.** Brief clinical report: the Nager syndrome. Am J Med General 1987; 27: 965–9.

[5] **Halal F, Herrmann J, Pallister P, Opitz JM, Desgranges MF, Grenier G.** Differential diagnosis of Nager acrofacial dysostosis syndrome: report of four patients with Nager syndrome and discussion of other related syndromes. Am J Med Gen 1983; 14: 209–24.

[6] **Butler MG, Hayes BG, Hathaway MM, Begleiter ML Specific genetic diseases at risk for sedation/anesthesia complications.** Anesth Analg 2000; 91: 837–55.

[7] **Vener DF, Lerman J.** The pediatric airway and associated syndromes. Anesthesiol Clin North Am 1995; 13: 585–614.

© 2015 Acta Medica. All rights reserved.