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Case Report Open Access

Ambulatory Anaesthesia in a Patient with Niemann-Pick Disease Type C

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Abstract

Niemann-Pick disease type C is an autosomal recessive lysosomal storage disorder that is characterized by severe visceral and neurologic involvement. The age of clinical onset diverges widely and the patients can present with a wide-ranging spectrum of signs and symptoms.

As literature on anaesthetic implications of these patients is scarce, we analysed the anaesthetic management of a 20-year old patient in a terminal progression state of the disease. The patient was admitted at our hospital for elective bilateral submandibular sialoadenectomy owing to chronic sialorrhea.

The difficulties and challenges arising during the anaesthetic procedure are discussed and a comprehensive analysis regarding the decision of discharging the patient at the same day of the procedure is also performed.

The main important directives for safe anesthetic management in Niemann-Pick disease type C patients are indicated with particular relevance being attributed to the anticipation and evaluation of each case individually.

Keywords: Ambulatory care; Anaesthesia, General; Niemann-Pick disease; Type C

Introduction

Niemann-Pick disease type C (NP-C) comprises alterations in the cellular transport of endocytosed cholesterol and accumulation of unsterified cholesterol in lysosomes and endosomes due to mutations of either the NPC1 or the NPC2 genes. The incidence of NP-C is approximately 1:120000 live births and most families (about 95%) have the NPC1 mutation [1].

NP-C is a neurovisceral condition with a wide range clinical spectrum, extending from a neonatal rapidly fatal disorder to an adult onset chronic neurodegenerative disease. The visceral involvement (liver, spleen and lung) and neurologic or psychiatric manifestations arise at different times and follow independent courses. When systemic disease is present, it always precedes the onset of neurological symptoms. The neurologic disorders consist of cerebellar ataxia, seizures, dysarthria, dysphagia, dystonia, cataplexy and progressive dementia. The majority of cases show vertical supranuclear gaze palsy and psychiatric disorders are common in late-onset patients. The observation of severe systemic impairment is unusual, excluding the perinatal period and lung involvement is not frequent [1,2].

Attending the complexity of NP-C progression, it is expected that these patients will require diagnostic or therapeutic procedures which comprise anaesthetic techniques. Nevertheless, literature on anaesthetic implications on NP-C patients is scarce and few cases have been published [2-4]. Hence, this case-report describes the anaesthetic management of a NP-C patient in a terminal progression state.

Case Report

A 20 years old female with NP-C (NPC1 mutation) was admitted at our hospital for elective bilateral submandibular sialoadenectomy owing to chronic sialorrhea.

The NP-C diagnosis was performed at age 11, although the first symptoms, including speech impairment and loss of cognitive skills, started at 8 year-old. Since then, the disease showed a progressive deterioration affecting the central nervous system and pulmonary functions.

On preoperative assessment, the patient has shown full dependence while performing daily living activitie and clinical neurological manifestations, including dystonia, dysphagia, seizures, aphasia and dementia, were observed. Simultaneously, the patient revealed a great difficulty in the elimination of salivary secretions, either because of dysphagia or decreased thoracic muscle strength. The accumulation of these secretions triggered apnea episodes and consequently airway permeability was supported by a cough-assist therapy and a suction device. Because of dysphagia and recurrent episodes of aspiration, it had been already performed a percutaneous endoscopic gastrostomy (PEG), which was achieved uneventfully under sedation.

At hospital admission, the patient weighed 40 Kg and the physical examination revealed an increased work of breathing and widespread ronchi over both lungs throughout pulmonary auscultation. Baseline saturation level without supplemental oxygen was 98% and the airway evaluation revealed no indicators of difficult ventilation or difficult intubation. Laboratory data was within the normal limits, with the exception of platelet number (1.19×10^9/L). No premedication has been prescribed.

Vital signs		
	Average	**CV**
MAP (mmHg)	65	0.15
HR (bpm)	92	0.11
SpO_2 (%)	98	0
MAP: Mean Arterial Pressure; HR: Heart Rate; SpO_2: Peripheral O_2 Saturation; CV: Coefficient of Variation		

Table 1: Vital signs during the anesthetic procedure.

In the operating room, the ASA (American Society of Anaesthesiology) standard monitoring, bispectral index (BIS) analysis and train-of-4 stimulation were applied. A 20-gauge abbocath was inserted with an infusion of a balanced electrolyte solution at 100 ml/h.

A smooth intravenous induction was achieved using remifentanil, 0.1 μg/Kg/min, propofol, 70 mg, and rocuronium, 10 mg. Mask ventilation was performed without difficulties and orotracheal intubation was completed by direct laryngoscopy using a 6.5 mm oral RAE (right angle endotracheal) tube. A Cormack-Lehane grade I view was obtained and the tube was positioned easily at the first attempt.

Anaesthesia was maintained using desflurane (3-5%), remifentanil (0.1-0.5 μg/Kg/min) and rocuronium (5 mg). Mechanical ventilation was provided at a tidal volume of 7 ml/Kg of predicted body weight in order to maintain low peak inspiratory pressure of up to 25 mmHg throughout the case. The respiratory rate was titrated between 14-16/min to achieve an $EtCO_2$ around 40 mmHg and the fraction of inspired oxygen was adjusted to keep target oximetry range higher than 95%.

Vital signs have been stable during the entire procedure (Table 1) and blood loss was estimated to be less than 50 ml. Ondansetron and paracetamol were administered, respectively, for postoperative nausea/vomiting prophylaxis and analgesia.

At the end of the procedure, all anaesthetic agents were discontinued and a bolus dose of sugamadex (100 mg) was administered. Extubation was performed considering the respiratory mechanics adequacy and after an adequate reversal of neuromuscular blockade (TOF ratio ≥ 90%).

The time elapsed between the discontinuing of anaesthetic agents and the arrival at the postanaesthetic care unit (PACU) was nearly 30 minutes.

Early recovery in the PACU was uneventful and when the patient has achieved a modified Aldrete score 5 of 9, after approximately 3 hours, she was discharged to a step down unit.

Considering the nature of surgery, the type of anaesthesia, the status of coexisting disease, the capabilities of outpatient facility and the dynamics and clinical evolution of the patient during PACU stay, it was decided to discharge patient home on the same day after phase II recovery. A postanaesthesia discharge scoring system (PADSS) [5] was applied to objectively assess the suitability for discharge. As a consequence, in the presence of stable vital signals, activity level consistent with preoperative baseline and the absence of pain, nausea, vomiting or signs of bleeding (PADSS 9), the patient was discharged home.

Discussion

NPD type C is a multisystem disorder which conveys simultaneously damage of the nervous system, alterations in respiratory system and hepatosplenomegaly. Consequently, several difficulties and challenges arise during the anaesthetic procedure. One of the upmost problems is related with lung ventilation, comprising complications associated with restrictive alterations in the lungs. Precision with ventilator parameters, namely higher respiratory rates and smaller tidal volumes, are mandatory in order to decrease the risk of barotrauma. In patients with liver disease, selection of the anaesthetic technique should be carefully considered as liver damage may change the metabolism of drugs.

Since the patient has shown no signs of liver disease, the main anaesthetic considerations were related to pulmonary disease and lung ventilation. We looked for a technique that would allow for rapidly and safely establishing satisfactory conditions for the performance of therapeutic procedure while ensuring rapid and predictable recovery with minimal postoperative sequel, such as the requirement of mechanical ventilation and intensive care admission. To attain this purpose, we decided to use remifentanil as an analgesic and desflurane for anaesthesia maintenance, since remifentanil has a reliable short context-sensitive half-life [6] and desflurane is rapidly washed-out after discontinuing because of its low solubility in blood and other body tissues [7]. For the same reason sugammadex was used at the end of the procedure to reversal neuromuscular blockade. However, in relation to desflurane it is also important to note that despite previous studies has been shown that it may irritate the respiratory tract at concentrations that exceed the minimum alveolar anaesthetic concentration (MAC), other studies revealed that respiratory complications that arise during maintenance delivery of anaesthesia are in agreement with the use of other inhalation anaesthetic agents [8]. Additionally, desflurane induces minimal cerebral cortical spike activity, which is also valuable in a patient with difficult-to-control epilepsy. Still, when contemplating the risks and advantages of desflurane utilization, we considered that a rapidly recovery would be fundamental in the anaesthetic management of this patient.

Despite the presence of preoperative risk factors associated with postoperative pulmonary complications [9], the surgical and anaesthetic techniques undergone without intercurrences or complications. The decision to discharge the patient on the same day of the surgery was discussed between the anaesthetic team, the surgical team and the patient mother and objective discharge scores were calculated concomitantly. The main advantages of an ambulatory procedure are associated with an earlier return to premorbid

physiologic state, with fewer complications and with reduced mental and physical disability [5].

In conclusion, NPD type C has important implications for safe anaesthesia management. Once possible comorbidities are already defined, it is imperative to anticipate, individually for each patient, the potential risk issues associated with perianaesthesia morbidity.

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