International Journal of Anaesthesia & Research
IJAR, 2(1): 26-28
www.scitcentral.com

ISSN: 2641-399X

Mini Review: Open Access

Perioperative Management of Patients with Lesch-Nyhan Syndrome

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Received March 06, 2019; Accepted March 11, 2019; Published April 06, 2019

ABSTRACT

Lesch-nyhan syndrome is associated with neurological, cognitive and behavioral disturbances. This syndrome has several problems such as positioning and difficult intravenous cannulation. These patients are prone for bradycardia, pulmonary aspiration, convulsion and sudden death. Anesthetic implications are not well described in these patients due to the low incidence of the disease.

This article's objective is to systematically review these needs in the perioperative period and how to approach them.

To conclude, patients with Lesch-nyhan syndrome should be carefully evaluated in the perioperative period and vigilant monitoring is required.

INTRODUCTION

Lesch-Nyhan syndrome is a rare X-linked recessive disorder with a prevalence of 1:380000 caused by the deficiency of hypoxanthine-guanine phosphoribosyltransferase (HGPRT) enzyme resulting in overproduction of purine and the accumulation of uric acid [1]. The syndrome almost always affects males, with very few cases reported in females [2]. The clinical features of Lesch Nyhan syndrome depends on whether there is complete or partial HPRT deficiency resulting in classical LND syndrome or a phenotype with some features absent or attenuated. Collectively, these patients are labeled as LND variants [3].

The classical clinical manifestations are characterized by neurological, cognitive and behavioral disturbances, congenital insensitivity to pain and uric acid overproduction. The most common presenting feature is developmental delay during the first year of life, with hypotonia and delayed motor skills usually evident by three to six months of age [1]. Affected children develop action dystonia (classical feature), choreoathetosis, opisthotonos and sometimes ballismus [4]. They also develop signs of pyramidal involvement including spasticity, hyperreflexia and extensor plantar reflexes [1]. Almost all affected individuals develop persistent self-injurious behavior between 1 to 3 years of age, a hallmark of the disease [5]. Self-injury most often involves biting of the fingers, hands, lips and cheeks [6]. Other compulsive behaviors may include aggressiveness, vomiting, spitting and coprolalia [1]. Hyperuricemia is associated with nephropathy, urinary tract calculi, arthritis, tophi. So these children may present for urinary tract, orthopedic surgeries or any unrelated surgical problems [2]. EEG may show nonspecific changes of slowing or disorganization.

There will be nonspecific changes of atrophy in the central nervous system with reduced cerebral volume and reduced caudate nucleus volume in CT and MRI scans. The findings of MRI revealed reduced basal ganglia volume which is consistent with the dystonic movement disorder [7].

METHODS

A search was performed on PubMed and Cochrane database using the following keyword combinations "lesch-nyhan syndrome", "anesthesia" and "surgery", "preoperative", "postoperative". 14 articles were selected from this revision. Inclusion criteria of the study were written in English, a systematic revision, case reports and case study. Only pediatric population was accepted in the group. Relevant papers cited in the selected article were added to the group ending up with 20 articles being analysed in this review.

ANESTHETIC IMPLICATIONS

The anesthetic considerations in Lesch-Nyhan syndrome are because of the functional disturbances created by the

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Citation: Sheetal K & Rachana Reddy AG. (2019) Perioperative Management of Patients with Lesch-Nyhan Syndrome. Int J Anaesth Res, 2(1): 26-28.

disorder and the effect on metabolism and excretion of drugs [2].

The main anesthetic concerns are:

1. Difficult Intravenous access
2. Difficult airway
3. Positioning
4. Risk of aspiration
5. Bradycardia, apnea and sudden death

Preoperative evaluation should be done with thorough history and examination to assess self-mutilating behavior, aggressiveness, developmental milestones, previous anesthetic exposure, cognitive dysfunction, difficult airway, spasticity, venous access, heart rate, blood pressure, sensory and motor functions, power and reflexes. Any other associated gastrointestinal disorders, respiratory, renal impairment, seizures, macrocytic anemia, malnourishment, arthritis, gouty tophi should be noted. Laboratory investigations like complete blood count to look for megaloblastic anemia, serum electrolytes, renal function tests and serum uric acid levels and ultrasound examination to rule out renal abnormalities, ECG to look for rhythm disturbances, chest x-ray are needed.

These children may present for urinary tract, orthopedic surgeries, dental extraction or any unrelated surgical problems. There are very few case reports of anaesthesia for a case of lesch-nyhan syndrome due to its rare incidence.

The patients with Lesch-Nyhan Syndrome have several problems like positioning in operating room, difficult intravenous cannulation and difficult airway because of the spasticitiy [8]. Also, these patients are at risk of bradycardia, pulmonary aspiration, convulsion and sudden death, and an increased incidence of vomiting.

The choice of anesthetic agents depends on the preoperative assessment, surgical problem, degree of metabolic dysfunction and renal function. Patients should be premedicated with metoclopramide to enhance gastric emptying and also use of H_2 receptor blocker to increase the gastric pH to 2.5 as the children are more prone for vomiting and aspiration pneumonia due to athetoid dysphagia which is characteristic in the syndrome [8].

Benzodiazapine like midazolam (oral or intravenous) which is most commonly used in pediatrics can be given to premedicate the child before induction to alleviate the anxiety [9]. The metabolisms of thiopental, ketamine or etomidate are not affected by reduced HGPRT function. Propofol is the preferred induction agent in such children10. The advantages of propofol include the ease of titration to anesthetic depth, antiemetic properties of the drug reduces the incidence of vomiting and may help in preventing aspiration [11]. It also increases urinary uric acid excretion and is beneficial for such patients [12]. Metabolism of most of the commonly used inhalational agents is not affected. Isoflurane is the preferred agent due to absence of nephrotoxicity [13]. Succinylcholine is preferably avoided because of abnormal potassium influx due to spastic muscle disorder [14]. Atracurium is safe for muscle relaxation [15]. Abnormalities of adrenergic responses, reduced monoamine oxidase activity have been reported [2]. The potential for bradycardia and heart block exists during the induction of anesthesia with propofol in any patient. Synergy of this reaction with the cholinergic predominance of the patient with LNS during stress may occur due to the absence of adrenergic response to stress [16,17]. Exogenous catecholamine should be administered carefully. A case report published by Reddy et al. [9] reports that there was decreased anesthetic requirement throughout the procedure and there was delayed recovery for 15 min. In this case, child had episodes of apnea during recovery when there were occasional ectopics and bradycardia which responded to injection atropine. So careful titration of drugs and vigilant intraoperative monitoring is required to prevent intraoperative complications.

The self-mutilation causes trauma to perioral tissues and subsequent scarring may suggest difficulties in endotracheal intubation [8]. A case report published by Salhotra et al. [18] reports the rare occurrence of a tracheal diverticulum associated with Lesch-Nyhan syndrome in an 11 year old boy. During induction of general anesthesia, unexpected difficult intubation was encountered with a 6.5 mm ID endotracheal tube and successively smaller tubes, also failing to pass 1 cm beyond the vocal cords. Intubation was finally successful with a 4.5 mm ID tube [18]. Difficult airway equipment and smaller size endotracheal tubes should be kept ready for such cases. Intraoperative reduced requirement of analgesics have been noted in these cases due to congenital insensitivity to pain [9].

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 [19]. Careful positioning of the patient with proper supporting can reduce, but may not be eliminate injuries to peripheral nerves [20]. Anesthesiologist should be familiar with the anatomical traces of the peripheral nerves to prevent pressure injury during the surgical procedure.

During extubation and recovery, these patients should be extubated after they have adequate spontaneous respiration and awake extubation is preferred due to difficult airway and monitored in the postoperative recovery till complete recovery from anesthesia as these children are more prone for respiratory compromise, convulsions and sudden death.

CONCLUSION

In conclusion, preoperative evaluation should be done to look for airway difficulty, body posture, difficulty in positioning, venous access, behavior, degree of developmental delay and other associated abnormalities. Anesthesia should be administered in slow and titrated doses. Necessary precautions should be taken to prevent vomiting and aspiration. Careful positioning with padding of the pressure points to prevent pressure injuries. Vigilant intraoperative as well as postoperative monitoring in the recovery as these children is more prone for convulsions, respiratory compromise and sudden death. Even though there are many contraindications to the standard anesthetic techniques for children with Lesch-Nyhan syndrome, anesthetic management should be individualized depending on the associated abnormalities and taking the basic precautions.

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