Introduction

Lesch-Nyhan syndrome is a rare, sex linked recessive anomaly consisting of a deficiency in the production of hypoxanthine phosphoribosyl transferase (HGPRT) enzyme. The patients with Lesch-Nyhan Syndrome will have spasticity due to which there could be several problems such as positioning, difficult intravenous cannulation and difficult airway. And these patients will have self-mutilating behavior and lack of pain sensation. Bradycardia, pulmonary aspiration, convolution are the important causes of death. Anesthetic management of these patients in pediatric age group is not well described. Here, we report our anesthesia experience in a 3 years-old child who was posted for debridement of right great toe with Lesch-Nyhan syndrome. Patients with Lesch-Nyhan syndrome should be induced in titrated doses and no analgesia is required because of lack of sensation. Careful monitoring is required during perioperative and post-operative period as they are more prone for bradycardia, apnea and aspiration.

Keywords: Anesthetic management; Lesch-Nyhan syndrome

Abstract

Lesch-Nyhan syndrome is a rare X-linked recessive disorder caused by a deficiency of Hypoxanthine-Guanine Phosphoribosyl Transferase (HGPRT) enzyme. The patients with Lesch-Nyhan Syndrome will have spasticity due to which there could be several problems such as positioning, difficult intravenous cannulation and difficult airway. And these patients will have self-mutilating behavior and lack of pain sensation. Bradycardia, pulmonary aspiration, convolution are the important causes of death. Anesthetic management of these patients in pediatric age group is not well described. Here, we report our anesthesia experience in a 3 years-old child who was posted for debridement of right great toe with Lesch-Nyhan syndrome. Patients with Lesch-Nyhan syndrome should be induced in titrated doses and no analgesia is required because of lack of sensation. Careful monitoring is required during perioperative and post-operative period as they are more prone for bradycardia, apnea and aspiration.

Case Report

A 3 year old boy weighing 15kg was scheduled for debridement of right great toe under general anesthesia. Child had a history of self-mutilating behavior of biting fingers, multiple injuries due to lack of sensation, delayed development, impaired sensation and speech disorder (Figure 1). Preoperative airway evaluation of the child revealed micrognathia, short neck, absent teeth and large glossy tongue (Figure 2). Laboratory test reports were as follows: Hemoglobin-12.4gm/dl, platelets-1.8lakh, Total count-6800, Serum creatinine-1mg/dl, blood urea-15mg/dl, serum uric acid levels were raised to 10.2 mg/dl. Serum electrolytes were in the normal range. Chest x-ray was normal. There was no cardiac or renal defect in this case.

The above physical findings predicted a difficult airway management. Therefore, before anesthesia induction all equipments were organized for difficult airway management. Child was premedicated with oral midazolam 0.5mg/kg in the recovery room 30minutes before induction. Child was shifted to operation theatre and all the monitors connected. Baseline recording of heart rate, blood pressure,
Lesch-Nyhan syndrome have several problems like positioning in operating room, difficult intravenous cannulation and difficult airway because of the spasticity [4]. Also, these patients are at risk of bradycardia, pulmonary aspiration, convulsion and sudden death, and an increased incidence of vomiting. The anesthetic considerations in Lesch–Nyhan syndrome are because of the functional disturbances created by the disorder and the effect on metabolism and excretion of drugs [2].

In our case we used propofol because of its antiemetic properties. It also increases urine uric acid excretion and is beneficial for such patients [5]. These children are more prone to nausea, vomiting [2]. Metabolism of barbiturates, ketamine, or etomidate remains unaffected and isoflurane can also be safely used [6]. Succinylcholine is preferably avoided because of abnormal potassium influx [7]. Atracurium is safe for muscle relaxation [8]. These children will have congenital insensitivity to pain [9]. There was no response to pain in this case. There was no postoperative pain. Perioperative proper positioning, use of protective padding, and avoidance of contact with hard surfaces are the protective measures to avoid direct pressure on susceptible skin and peripheral nerves [10].

In our case, there was decreased anaesthetic requirement throughout the procedure. There was delayed recovery for 15min. Child was intermittently going into apnea during which there were occasional ectopics and bradycardia which responded to injection atropine. These children are more prone to bradycardia and apnea so vigilant monitoring and intervention is required for such cases [2].

A case report published by Salhotra R et al., reports the rare occurrence of a tracheal diverticulum associated with Lesch-Nyhan syndrome in a 11 year old boy [11]. They had difficulty in securing the airway.

In conclusion, thorough preoperative evaluation of the child to assess the airway, cannulation and any associated abnormalities. Induction should be done with titrated doses. Precautions should be taken to avoid aspiration. Careful monitoring throughout the surgery to prevent complications and sudden death. No analgesics required due to lack of sensation in these patients. Patients should be closely monitored in the postoperative period because of the risk of sudden death and aspiration. Anaesthetic management should be individualized for each case of Lesch-Nyhan syndrome based on the associated conditions.

References


Discussion

Lesch-Nyhan syndrome is characterized by cognitive impairment, hypotonia at rest, choreoathetosis, hyperuricaemia and the hallmark symptom of severe and involuntary self-mutilation. The patients with Lesch-Nyhan Syndrome have several problems like positioning in


