Case Report

An unanticipated difficult airway in Lesch–Nyhan syndrome

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Abstract

An 11-year-old boy with Lesch–Nyhan syndrome presented to the emergency for fixation of a fractured femur. 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 achieved with a 4.5-mm ID tube. The surgery was completed uneventfully. A tracheal diverticulum was found in the computerized tomography (CT) scan performed postoperatively to account for this unexpected difficult intubation. This case highlights the anesthetic concerns in Lesch–Nyhan syndrome and also reports the rare occurrence of a tracheal diverticulum associated with it.

Key words: Difficult airway, Lesch–Nyhan syndrome, tracheal diverticulum

Introduction

Lesch–Nyhan syndrome is a rare disorder of purine metabolism caused by deficiency of hypoxanthine-guanine phosphoribosyltransferase (HGPRT), produced by mutations in the HPRT gene. While renal dysfunction, musculoskeletal abnormalities, and impaired drug metabolism are previously documented concerns,[1,2] there is no evidence to suggest difficulty with intubation. This case highlights the presence of difficult intubation associated with Lesch–Nyhan syndrome. The cause was ascertained postoperatively to be a tracheal diverticulum, a rare entity with an incidence of 0.25–1%.[3]

Case Report

An 11-year-old boy presented to the operation theater for emergency fixation of fractured femur. He was recently diagnosed to have Lesch–Nyhan syndrome, with its most striking feature of self-mutilation seen in the right thumb and lower lip [Figure 1]. The patient was conscious and responding to verbal commands, had evidence of mental retardation, spasticity, and hyperuricemia (serum uric acid levels: 12.5 mg%, normal range: 3.6–8.3 mg%), albeit no nephropathy was present (serum creatinine: 0.9 mg%). Upper respiratory tract infection along with stridor was also noted.

General anesthesia was planned considering the presence of mental retardation and spasticity. Due to the inspiratory stridor, a difficult intubation cart including a cricothyrodotomy set was kept ready. The SpO_2 on room air was 96–98%. Following institution of routine monitoring, inhalation induction was done with sevoflurane 4–5% in oxygen. After ensuring successful manual ventilation, muscle relaxation was achieved with 15 mg atracurium. On laryngoscopy, the aryepiglottic folds were edematous and congested with a view of Cormack-Lehane grade II. Intubation was attempted with a

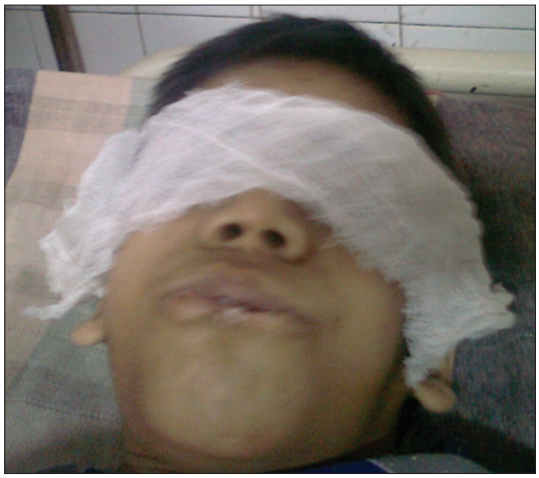

Figure 1: Self-mutilation of the lips

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6.5 mm ID cuffed endotracheal tube, and although it passed the vocal cords with ease it could not be advanced 1 cm beyond the vocal cords. Intubation with progressively smaller sized tubes, i.e., 6.0, 5.5, and 5.0 mm ID was attempted, but all of them failed to pass 1 cm beyond the vocal cords. Finally, a 4.5 mm ID sized uncuffed tube could be successfully advanced and was used to secure the airway.

Rest of the intraoperative and postoperative course was uneventful. Following reversal of neuromuscular blockade, though the oxygenation was not compromised and the patient regained consciousness, he had stridor. Considering the edema viewed on laryngoscopy, nebulisation with budesonide 1 mg stat followed by 0.5 mg 8 hourly was started.

In the postoperative period, a CT scan of the tracheobronchial tree was done to diagnose the cause for difficulty in intubation. The scan revealed a tracheal diverticulum 9 mm below the vocal cords on the left posterolateral wall of the trachea [Figures 2a and b]. It was 2.5 cm deep and had a wide mouth (1.7 cm). The expected dimensions of the trachea for this age are 1.16 ± 0.1 cm anteroposteriorly and 1.18 ± 0.09 cm transversely.[4] The cross section of trachea was significantly decreased 3.5 cm below the cords and distal to the diverticulum, with the narrowest diameter being 0.5 cm [Figure 2a].

Discussion

Encountering a case of Lesch–Nyhan syndrome for the conduct of anesthesia is rare. In our patient, it was associated with an unanticipated difficulty in securing the airway. There was no limitation in mouth opening or difficulty in introduction of laryngoscope, although perioral trauma due to self mutilation is hypothesized to interfere with mouth opening.[5] Negotiating progressively smaller sized (6.5, 6.0, 5.5, and 5.0 mm ID) endotracheal tubes, 1 cm beyond the vocal cords, was difficult despite an easy passage through the cords. Subglottic obstruction has been reported to result from subglottic stenosis,[6] laryngotracheobronchitis, laryngotracheitis,[7] post intubation laryngeal stenosis,[8] and tracheobronchopathia osteoplastica.[9] There is only one case report of tracheal diverticulum resulting in difficulty with intubation, where the endotracheal tube could not be passed 2 cm beyond the cords.[10] The mouth of diverticulum, present 9 mm below the cords on the posterolateral wall of trachea, was the probable site of impaction of endotracheal tubes [Figure 2a]. A 4.5-mm ID tube has a smaller radius of curvature than larger tubes [Figure 3]. The "more curved" 4.5 mm tube probably avoided impinging on the tracheal diverticulum and thus passed into the trachea successfully.

Tracheal diverticulum may be congenital or acquired in origin.[10] Congenital diverticula are small and narrow-mouthed and may occur in isolation or associated with other congenital abnormalities in the tracheobronchial tree. Acquired diverticula are thought to be out-bulgings from weak areas in the posterior tracheal wall. They are typically wide-mouthed and larger than the congenital ones, as seen in our patient. Secretions can pool in them and cause infection of the tracheobronchial tree.

On most occasions tracheal diverticulum is an incidental finding on autopsy, bronchography, CT scan, or endoscopy.[10] It may remain asymptomatic throughout life or present with chronic cough or respiratory infection,[11] hoarseness of voice, respiratory distress,[12] recurrent laryngeal nerve palsy,[13] or a neck swelling.[12] There is a case report of ventilatory difficulty after tracheal intubation in a patient with a tracheal diverticulum[14] and another case report of pneumomediastinum secondary to accidental perforation of a diverticulum caused by tracheal intubation.[15] When encountering an obstruction to endotracheal tube passage, forceful advancement may lead to such complications.

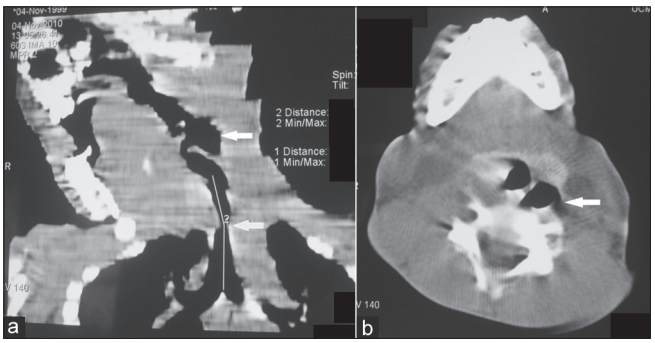

Figure 2: (a) Diverticulum (white arrow) and narrowest part (yellow arrow) of the trachea; (b) Diverticulum (white arrow) alongside the trachea

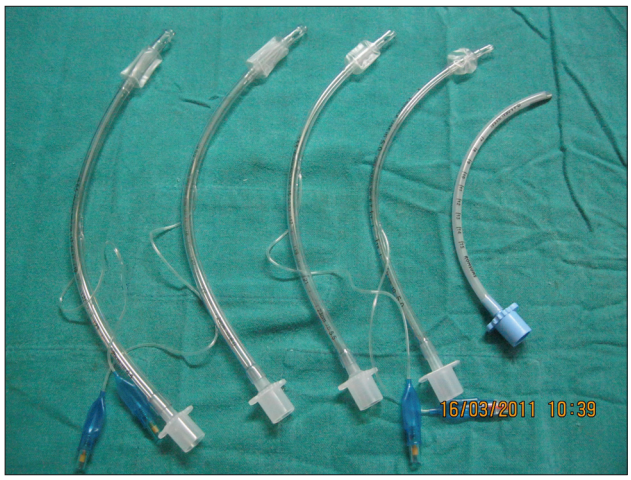

Figure 3: Tube curvature comparisons: 6.5, 6.0, 5.5, 5.0, and 4.5 mm ID from left to right

Treatment for tracheal diverticulum may be surgical or conservative. No active management needs to be done in asymptomatic cases.[11] In chronically symptomatic cases with infection of the tracheobronchial tree antibiotics, mucolytics, bronchodilators, postural drainage, and physiotherapy are the mainstay of management.[11,16] Surgical resection, although rare, can be considered for children.[13,16,17] Our patient refused any further investigation or surgical interventions.

Our patient had inspiratory stridor during the perioperative period possibly due to supraglottic edema, which continued till he was discharged, after 30 days of hospitalization. The stridor persisted despite the inhaled steroids and antibiotics, though it decreased in severity. Tracheal diverticulum is known to cause respiratory distress and stridor by spillage of secretions collecting in the diverticulum into the trachea[12] and this may explain the persistent stridor in our patient. However, it was never associated with altered consciousness or desaturation.

Lesch–Nyhan syndrome is an X-linked recessive disorder of purine metabolites leading to hyperuricemia and hyperuricosuria with consequent nephropathy, and neurological effects such as poor muscle control, choreoathetosis, spasticity, self-mutilation, and mental retardation.[1] Lesch–Nyhan syndrome almost always affects males, with very few cases reported in females. [1] Our patient presented with the typical manifestations of self-mutilation, mental retardation, spasticity, and hyperuricemia, but there was no nephropathy. Postoperatively, he responded to allopurinol therapy (100 mg twice a day) with fall in serum uric acid levels (6.8 mg% after 2 weeks of therapy).

The anesthetic considerations in Lesch–Nyhan syndrome are consequent to the functional disturbances created by the disorder and the effect on metabolism and excretion of drugs.[2] Spasticity leads to difficulty in patient positioning and securing intravenous access,[5] while athetoid dysphagia makes these patients prone to aspiration pneumonia.[5] Hyperuricemia may lead to urinary calculi and nephropathy. Drugs metabolized by the kidneys are preferably avoided. Metabolism of barbiturates, ketamine, or etomidate remains unaffected and isoflurane can also be safely used.[18] Propofol increases urine uric acid excretion and is beneficial for such patients.[19] Succinylcholine is preferably avoided because of abnormal potassium influx.[20] Atracurium is safe for muscle relaxation.[21] Abnormal and complex adrenergic responses have been reported in patients with Lesch–Nyhan syndrome, mandating an extra care when administering exogenous catecholamines.[2]

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