A rare presentation of a child with osteogenesis imperfecta and congenital laryngomalacia for herniotomy

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ABSTRACT

Sometimes anaesthesiologists come across rare congenital anomalies in their practice. The inherent complications associated with the disorder necessitate tailor-made approaches for providing anaesthesia to even seemingly simple surgical interventions. Here, we share our experience of anaesthesia management of an infant with congenital laryngomalacia and recently diagnosed osteogenesis imperfecta type 1 who had presented to us with an acute abdomen for a semi-emergency herniotomy.

Key words: Congenital laryngomalacia, general anaesthesia, osteogenesis imperfecta, paediatric

INTRODUCTION

Laryngomalacia is the most common congenital anomaly of the larynx and the commonest cause of stridor of unknown aetiology in infants, and accounts for around 75% of all laryngeal problems in infancy.[1]

Osteogenesis imperfecta is a rare congenital anomaly of the connective tissues caused by the mutation of collagen type 1, COLIA1 or COLIA2 genes. It affects the bones, sclera and the inner ear and has an incidence of about 1 in 50,000.[2]

The co-existence of these diseases is rare and it presents unique anatomical and physiological challenges to the anaesthesiologist making every form of anaesthesia difficult.

CASE REPORT

A 6-month-old male infant, 3.5 kg in weight, presented with the history of refusal to feed, tender abdomen, swelling in the right groin and constipation since 3 days. Diagnosis of a right-sided obstructed, irreducible indirect inguinal hernia was made. The paediatric surgeons reduced the hernia and a semi-elective herniotomy was planned.

The child had a history of stridorous breathing since birth and was subsequently diagnosed as a case of congenital laryngomalacia. He was a full-term delivery by Caesarean section for meconium-stained liquor. The infant was kept in the intensive care unit (ICU) for 3 days post-delivery for breathing difficulties and neonatal jaundice. Anaesthesia pre-evaluation revealed microretrogenathia [Figure 1]. The child underwent a High resolution computed tomography (HRCT) imaging of the chest along with virtual bronchoscopy to rule out any obstruction in the airway and there was a chance finding of three ununited rib fractures of the left side (4th, 5th and 6th) [Figure 2]. Subsequent paediatric, orthopaedic and ophthalmologic evaluations revealed blue sclera, spontaneous rib fractures, failure to thrive and multiple hernias (umbilical and right inguinal) because of the lax...
abdominal wall. Cardiac evaluation was unremarkable. A clinical diagnosis of congenital osteogenesis imperfecta type 1 was made by the paediatricians.

High-risk consent and consent for post-operative ventilation was obtained. The child was premedicated with inj. glycopyrolate 4 mcg/kg, inj. midazolam 0.03 mg/kg and inj. ketamine 0.5 mg/kg IV. Anaesthesia was induced with IV propofol 2 mg/kg. A paediatric fibreoptic bronchoscope was not available, so direct laryngoscopy was performed with a shoulder pad in place and intubation was achieved in the second attempt using size 1 Miller blade, and a 3.5-mm uncuffed endotracheal tube (ETT) was inserted. On direct laryngoscopy, the view was Cormack–Lehane Grade III. After confirmation of tube placement, inj. atracurium 0.5 mg/kg was administered and anaesthesia was maintained with oxygen, nitrous oxide, and sevoflurane. Monitoring devices included a pulse oximeter, cardioscope, non-invasive blood pressure monitor, temperature monitor and capnometer. Caudal block was administered with 2 cc of 0.25% bupivacaine. The patient was carefully repositioned supine and adequate padding was done. The surgery lasted 30 min and was uneventful. Post-surgery, neuromuscular blockade was reversed and the patient extubated. The patient was shifted to the ICU for observation. A quiet and pain-free post-operative period ensured that the patient did not have any breathing difficulties.

**DISCUSSION**

Laryngomalacia is defined as severe collapse of the epiglottis and the arytenoids (as seen by fibreoptic bronchoscopy) in children with audible stridor. Congenital laryngomalacia presents in the neonatal period with stridor, that is high pitched in nature and inspiratory in timing and is aggravated when the child is upset or when the child hyperventilates. In more severe forms, upper airway obstruction, cyanotic spells, feeding difficulties, failure to thrive, cor pulmonale and developmental delay occur. The definitive diagnosis is made by suspension laryngoscopy and direct visualization of the larynx as it collapses during the inspiratory phase of spontaneous ventilation.

Osteogenesis imperfecta, also known as the brittle bone disease, is a genetic disorder caused due to the faulty formation of collagen type 1. The disease may cause cardiac valvular lesions, cor pulmonale, neurologic abnormalities, hyperhydrosis, cleft palate, metabolic abnormalities, malignant hyperthermia, obstructive uropathy and platelet dysfunction.\(^3\)\(^4\)

Mild cases of laryngomalacia may require only history, clinical examination, complete blood count and electrolytes, with lateral neck and chest X-rays. An electrocardiogram (ECG) may be ordered if there are signs of cor pulmonale. Pulmonary function tests are required in cases of kyphoscoliosis associated with osteogenesis imperfecta. Barium studies, chest CT scan and angiography are reserved for those cases where a cardiovascular anomaly is present.

Premedication of patients with laryngomalacia is essential to prevent vagal responses and as an antisyialogue, sedation is to be used judiciously. We opted for mild sedation to prevent the child from crying and hyperventilating as it would have worsened the stridor.

Laryngomalacia may make mask ventilation a challenge as the airway may collapse when the muscle tone is lost following induction. Facial anomalies associated with osteogenesis imperfecta like retrognathia, combined with a large head, short neck and propensity to mandibular fractures make mask ventilation difficult.
The intubation of patients with laryngomalacia is difficult due to the large lax overhanging epiglottis. Facial anomalies, risk of mandibular fractures, injury to teeth and high chances of cervical vertebrae injury associated with osteogenesis imperfecta make direct laryngoscopy and intubation difficult. Intubation with a paediatric fibreoptic bronchoscope would be the method of choice in these cases. The use of a laryngeal mask airway (LMA) in a case of laryngomalacia has a higher failure rate and it may not be able to protect the airway against reflux that is commonly seen in these patients.

Patients with osteogenesis imperfecta have a higher propensity to malignant hyperthermia as compared to the general population, but the incidence is low. In anticipation of a difficult airway, we proceeded with the use of sevoflurane with temperature and EtCO₂ monitors in place. Total intravenous anaesthesia would be the method of choice in cases where a difficult airway has been ruled out.

Other than triggering malignant hyperthermia in susceptible patients, succinylcholine-induced fasciculations can cause fractures and neck hyperextension leading to atlanto-occipital dislocations. It was therefore avoided in our case. Also thyroid storms can occur intraoperatively, as 50% of the patients with osteogenesis imperfecta have increased levels of thyroxine.

Patients should be positioned carefully, with adequate padding as fractures and dislocations are very common in osteogenesis imperfecta. In a patient given general anaesthesia, dislocations and fractures occurring intraoperatively may go unnoticed.

Patients of osteogenesis imperfecta have a tendency for bleeds in spite of having a normal coagulation profile and bleeding time. Coagulopathy with a sudden development of widespread petechiae has also been reported. Therefore, due precautions regarding any unexpected bleeding were taken in the form of availability of adequate blood, fresh frozen plasma and platelet concentrates.

Regional anaesthesia is the method of choice for patients with osteogenesis imperfecta in view of all the difficulties enumerated above, but when combined with laryngomalacia, securing the airway gains prime importance. When general anaesthesia is considered, meticulous attention is required with the use of neuromuscular blocking agents, inhalational agents, airway management, positioning of the patient and acute pain management.

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