Osteogenesis Imperfecta- An Anaesthesiologist’s Challenge

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**PRESENTATION OF CASE**

We present a case of 26 year male posted for shaft of femur fracture with history of osteogenesis imperfecta tarda type IV with severe anatomical deformities who underwent nailing procedure. Procedure was undertaken under general anaesthesia and epidural catheter was inserted postoperatively for management of pain. A 26-year old male presented with fracture shaft left femur and was planned for nailing procedure electively. He was a known case of osteogenesis imperfecta tarda type-IV with characteristic features of short stature, brittle bones, hypermobile joints, kyphoscoliosis and history of recurrent fractures of long bones for which he was operated previously twice under general anaesthesia, which was uneventful. Patient’s sister is also a patient of osteogenesis imperfecta tarda type IV.

On general examination he was observed to be short stature, (105 cm), 40 kg, afebrile with normal coloured sclera. His pulse and blood pressure were within normal limit. Respiratory system revealed barrel shaped chest with bilateral equal air entry. On airway assessment he had acceptable flexion and extension at neck with adequate mouth opening and normal dentition. Airway assessment was of Mallampati class IV. Kyphoscoliosis was seen on examination of spine. Systemic examination was normal.

**Figure 1**

**Figure 2**

**Figure 3**
CLINICAL DIAGNOSIS

26 year male with osteogenesis imperfecta tarda type-IV posted for nailing for fracture neck shaft of femur.

PATHOLOGICAL DISCUSSION

Osteogenesis imperfecta is an inherited autosomal disorder of connective tissue origin. It is most commonly known as a ‘brittle bone disease’ as these patients commonly present with recurrent fractures after trivial trauma. It is caused due to mutations in collagen type 1 COL1A1 or COL1A2 genes.

The disease is classically divided into two syndromes- Congenita and tarda where tarda is associated with normal life expectancy. Silence classified the disease into four distinct types. Type -II and Type -III are autosomal recessive. Type -I and Type-IV -both autosomal dominant. Autosomal dominant variant is characterized by short stature, bone fragility leading to frequent fractures and dentinogenesis resulting in easily broken teeth. The fractures in type -I, the most common of the four disorders are generally non-deforming and characteristic blue sclera while fractures in type-IV tend to cause deformities of long bones and thoracic cage.

The defect in skeletal growth is due to lack of normal ossification of enchondral bone which increases bone fragility. Hence, patients have history of recurrent fracture of bones and have hypermobile limbs and other associated skeletal deformities like kyphoscoliosis, short neck, pigeon chest with difficult airway and risk of odonto-axial dislocation of cervical vertebra, mandible and teeth fractures during laryngoscopy and intubation. The patients may also have other systemic abnormalities such as cardiac valvular lesions, cor-pulmonale, neurologic abnormalities, hyperhidrosis, cleft palate, metabolic abnormalities, malignant and non-malignant hyperthermia and obstructive uropathy following renal and ureteric stones and platelet dysfunction.

We hereby share our experience of anaesthetic management of the above mentioned case.

DISCUSSION OF MANAGEMENT

Routine haematological investigations were done including coagulation profile which were normal. Thyroid function test, liver function test, renal function test and creatine kinase were normal. Electrocardiogram and Echocardiogram were normal. Chest radiogram revealed normal cardiac shadow. X-ray spine revealed dorsolumbar kyphoscoliosis to left.

Patient was accepted for surgery with ASA grade III risk. In the operating room, the patient was carefully placed in the supine position, routine monitors were then applied (ECG, pulse oximeter, skin temperature probe). Blood pressure was measured manually. Intravenous line secured in left hand with 18 G canula. Total 1000 ml of Ringer lactate and 500 ml of DNS was given after prewarming to body temperature. Patient received premedication with ondansetron 4 mg, ranitidine 50 mg and preoxygenated with 100% O₂ for three minutes. Anaesthesia was induced with propofol 100 mg+ fentanyl 50 mcg and ability to mask ventilation was assessed before administering Rocuronium 40 mg. Then IPPV was done for three minutes and Endotracheal tube coughed 6.5 was inserted and ventilation was followed with Closed circuit on pressure-controlled ventilation mode. EtCO₂ monitoring was in place throughout the perioperative period. Anaesthesia was maintained using intermittent rocuronium with O₂/N₂O/Sevoflurane 1 MAC. For analgesia Diclofenac Aq. 75 mg was used intramuscularly. Right lateral position was required for the planned surgery. Meticulous care was taken when the patient was being shifted to right lateral position in the form of appropriate padding on pressure areas with preformed cushions & cotton pads. Intraoperative period of 2 hours remained uneventful. At the end of the surgery, epidural catheter 18 G was inserted using Tuohy needle at L3 L4 space for post-operative pain management. Patient was repositioned to supine and reversal of anaesthesia was carried out using 100% O₂ with neostigmine 2.5 mg and glycopyrrolate 0.4 mg. Endotracheal tube was removed after resumption of regular spontaneous respiration, but at a deeper plane of anaesthesia to prevent return of excessive muscle tone. Postoperative period was un-eventful, and patient was subsequently discharged on the seventh postoperative day.

Although our patient received general anaesthesia, the best anaesthetic technique in patients with Osteogenesis imperfecta is conduction block (regional anaesthesia) for various reasons, firstly, because it avoids the necessity for tracheal intubation (laryngoscopy and tracheal intubation associated with a risk of odontoaxial dislocation, fracture mandible, cervical vertebrae and injury to teeth in patients with osteogenesis imperfecta). Secondly, regional anaesthesia decreases the chances of patient developing hyperthermia compared to general anaesthesia (as malignant hyperthermia is the result of either an abnormal central nervous system temperature regulating mechanism or abnormal cellular energy metabolism). Lastly, it helps in easier detection of thyroid storm which is common in at least 50% of patients with this disease. In the above-mentioned case anatomic deformity is more prominent compared to the physiological deformity. General anaesthesia was the choice of anaesthesia opted due to the presence of dorso-lumbar kyphoscoliosis. Kyphoscoliosis can predispose these patients to inadvertent dural puncture and coupled with short stature, which makes it difficult to predict the level of block produced by a given dose of local anaesthetic. Karabiyk et al have recommended TIVA along with intubating LMA to manage the elective case. Sachin et al noted a significant degree of movement between first and second cervical vertebra (odontoaxial) during direct laryngoscopy and with the use of intubating LMA.
Difficult airway must always be anticipated in such patients, due to abnormal skeletal growth, short stature and hypermobile joints. Therefore, we were ready with difficult airway kit (including ILMA and fiberoptic device).

Succinylcholine induced fasciculations may cause fractures, as may hyperextension of neck and risk to trigger the malignant hyperthermia, hence it was avoided in this case. An automated arterial pressure cuff may be hazardous, as over-inflation can result in a fracture, therefore, we used a manual sphygmomanometer. Bleeding may occur despite normal results of coagulation studies and bleeding time, which makes predictions about intraoperative bleeding difficult. Coagulopathy with 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.

In our opinion, extra caution needs to be taken in patients of osteogenesis imperfecta undergoing operation in the lateral position and under general anaesthesia since the combination of muscle relaxants and the possibility of overlying weight of the upper body on the dependent shoulder in this patient further increased the chances of dislocation as well as fracture in view of their already lax joints and brittle bone. Further, the possibility of such problems remaining undetected in the unconscious, anaesthetized patient should be borne in mind. Standard auxiliary rules may be custom made for a patient in the lateral position, prior to induction of anaesthesia. Repeated intra operative checks of pulses will also help in excluding or detecting occurrence of any such events in the intraoperative period. After recovery from muscle relaxants with the analgesia and sedation any underlying problem associated with position e.g. pressure on neurovascular bundle may lead to neuropaxia which may remain undetected for a longer period and thus patient may have consequent damage resulting in lawsuits in consumer court for the damages.

Regional anaesthesia is the technique of choice in such cases, but when general anaesthesia is considered in view of proposed surgical procedure or due to relative contra indication of regional block, as in this case, meticulous attention is required especially with the use of neuromuscular blocking agents, inhalational agents, airway management, positioning of the patient and acute pain management.

**FINAL DIAGNOSIS**

26 years old male patient with fracture shaft of femur, known case of osteogenesis imperfecta posted for nailing managed under general anaesthesia with adequate precautions.

**REFERENCES**


