Anaesthetic Management of A Case of Osteogenesis Imperfecta with Urinary Bladder Stone - A Case Report

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Summary

Sometimes in practice of anaesthesia, anaesthesiologist encounters patients with rare congenital diseases. To anaesthesiologist, these patients are a challenge due to inherent complications associated with the disease. Here, we are reporting a case of osteogenesis imperfecta who was posted for the surgery for vesical calculus. All investigations were done to rule out any cardio-respiratory abnormalities, bleeding disorders, which are commonly associated with these patients. Caudal epidural was chosen as anaesthesia technique of choice as spinal anaesthesia was anticipated to be difficult due to associated kyphoscoliosis. GA was avoided due to anticipated difficult airway, restrictive lung disease and susceptibility to malignant hyperthermia. We emphasize the importance of proper preanaesthetic evaluation, intellectual, mental and logistical preparation which should be done before anaesthetising these types of patients.

Key words Osteogenesis Imperfecta, Bladder stone, Anaesthesia

Introduction

Osteogenesis imperfecta(OI), also known as brittle bone disease, is a genetic disorder of connective tissue characterized by bones that fracture easily, often with little or no trauma. Osteogenesis imperfecta is caused by a faulty gene that instruct to make too little or poor quality of type 1 collagen. The prevalence of osteogenesis imperfecta ranges from 1:60000 to 1:20000 depending upon type of OI. Inheritance in nearly all cases follows an autosomal dominance pattern, although sporadic cases are common. The disorder is frequently associated with blue sclera, dental abnormalities (dentinogenesis imperfecta), progressive hearing loss, and a positive family history. The most common classification for OI was developed by Sillence.

Anaesthetic implication of OI includes difficult intubation, platelet dysfunction, cardiovascular abnormalities like mitral valve prolapse, tendency to develop malignant hyperthermia and problems with positioning of patient due to brittle bones.

Case report

A 54-year-old male patient presented in emergency room with acute retention of urine. He was a known case of OI tarda (Fig 1). Immediately foley’s catheterization (no.-16fr.) was done to relieve retention. X-Ray KUB of the patient revealed a right side renal calculus and a vesicle calculus and was planned for cystolithotomy (Fig 2).

Fig 1 Photograph showing skeletal abnormalities
surgery, his vitals remained stable and no rescue medication was required. Operation lasted for about 40 min and patient had an uneventful recovery. He was discharged on 5th postoperative day.

Discussion

Osteogenesis imperfecta is a rare autosomal dominant inherited disease of connective tissues that affects bones, sclera and inner ear. The incidence is higher in females. Clinically, it occurs in two forms: osteogenesis imperfecta congenita and osteogenic imperfecta tarda. With congenital forms fractures occur in utero and death is usually in perinatal period. The tarda form typically manifests during childhood or early adolescence, but the patients have a normal lifespan.

Management of anaesthesia is influenced by co-existing orthopaedic deformities, vulnerabilities to fracture during perioperative period, associated cardiac abnormalities, impaired platelet function, tendency to develop hyperthermia and rarely extra skeletal manifestations. Due to abnormal skeletal growth difficult airway must always be anticipated in such patients. Associated kyphoscoliosis along with pectus carinatum may decrease vital capacity, chest wall compliance with resulting arterial hypoxemia due to ventilation perfusion mismatch and this can lead to increased risk under GA. Succinylcholine should be avoided as fasciculation can lead to fractures. Regional anaesthesia is acceptable in selected patients as it avoids need for tracheal intubation but may be difficult because of kyphoscoliosis. Before giving regional anaesthesia, coagulation profile must be screened due to associated increase in bleeding time despite normal platelet count.

For monitoring of blood pressure, automated blood pressure cuffs may be hazardous as over inflation may result in fracture. During prolonged surgery, all pressure points should be well padded and positioning of patient along with transportation should be very gentle to prevent occurrence of fracture.

There have been several successful case reports of conductance of surgery under general anaesthesia in...
patients with osteogenesis imperfecta. Karabiyik et al. have recommended TIVA along with ILMA to manage elective case, while Malde et al. have successfully used balanced general anaesthesia in a case of osteogenesis imperfecta with gross deformity of pelvis for abdominal hysterectomy.

In our patient, we avoided general anaesthesia due to anticipated difficult airway (attributed to limited mobility of cervical spine, short neck and absent dentition), restrictive lung disease (due to associated kyphoscoliosis, pectus carinatum) and susceptibility to malignant hyperthermia. However, preparation was kept ready in operation theatre for managing difficult airway in case of emergency.

We preferred regional anaesthesia since the patient had to undergo a lower abdominal surgery and to avoid risk related to general anaesthesia. Before giving regional anaesthesia, a thorough preoperative workup of patient was done with special attention to coagulation profile as these patients are prone to have abnormal bleeding tendencies. Patient’s BT, CT and PT were within normal limits. Caudal epidural was chosen as preferred anaesthesia technique over spinal anaesthesia as it was difficult to perform lumboperitoneal puncture due to associated kyphoscoliosis and unpredictability of the level of block. The effect of caudal block was till T10 level and course of surgery was uneventful.

To summarise, patients with OI pose a significant challenge to anaesthesiologist owing to difficult airway, problems with positioning, fractures, tendency for hyperthermia and platelet functional abnormalities. Only thorough preoperative workup and prompt management can improve the outcome in these patients.

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