Anaesthetic management in a patient with osteogenesis imperfecta and a fractured femur

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Introduction

Anaesthesiologists frequently encounter rare congenital diseases and syndromes. Osteogenesis imperfecta (OI) is a fibro-osseous disorder of the collagen tissue. OI is also known as brittle bone disease, and is a genetic disorder of connective tissue in which bones fracture very easily, often for no apparent reason. The aetiology of the disease is a gene defect that produces very little or poor quality type 1 collagen, an important building block of bones.[1] Usually, patients inherit the disease from a parent, but sometimes cases are sporadic and the result of new genetic mutation.[2]

Other clinical features of the disease include blue sclerae, progressive deafness, brittle teeth (dentinogenesis imperfecta) and hypermobile joints. In the severe form of the disease, kyphoscoliosis may lead to significant chest wall deformity and restrictive lung disease. These two pathological conditions, together with recurrent pneumonia, can progress to cardiac failure.[3] The central nervous system may be involved, with spinal cord or brainstem pressure effects, like basilar invagination or impression and craniovertebral junction problems.[4]

The incidence is 6-7 per 100 000 people, leading to defects in skeletal growth.[5] These patients present with short stature and are challenging to anaesthesiologists because of the multi-system organ involvement of OI. Most notably, anaesthesiologists are challenged by a difficult airway, the fragile skeletal system during positioning, cardiovascular involvement, coagulation abnormalities and an association with malignant hyperthermia.[2]

The diversity of presentation means that patients with severe forms may present with multiple fractures with minimal or no trauma, and persons with mild forms may only manifest with premature osteoporosis or severe postmenopausal bone mineral loss.[6] There is no cure for the disease, and currently medical management remains symptomatic. Intramedullary nailing of long bones remains the method of choice for correcting deformities and preventing fractures.[7]

We report on an OI patient with a fractured shaft of femur who underwent internal femur fixation. The perioperative management of this patient is discussed and the literature reviewed.

Case report

A 21-year-old female, weighing 48 kg, with a height of 140 cm, and a history of repeated fractures since the age of 10 years, presented to our centre after sustaining a fractured right femoral shaft associated with a fall (Figure 1). She was a known case of OI tarda. Our institution is a tertiary care centre that caters to the needs of the people of northern India. The patient's history revealed an uneventful antenatal period and no delay in milestones. Past surgical history was suggestive of a fractured shaft of the right femur six years ago, for which intramedullary nailing was completed under general anaesthesia (Figure 2). There was no family history of the disease. The patient was awake and alert at the preoperative assessment. A general physical examination revealed defective dentition, a short neck, kyphoscoliosis and valgus deformity bowing deformities in the lower limb (Figure 3). There was no pallor, icterus, cyanosis, clubbing, lymphadenopathy or pedal oedema. Her blood pressure was 124/70 mmHg and her heart rate 80 beats/minute. The O_2 saturation in the room air was 96%. The cardiac status and other systemic examinations of the patient were normal, except for the musculoskeletal examination, which revealed kyphoscoliosis and valgus deformity of the lower limb.

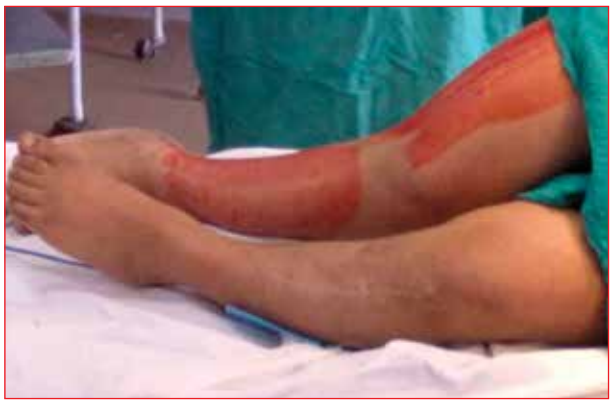

Figure 1: Showing the patient with bow deformity of the lower limbs

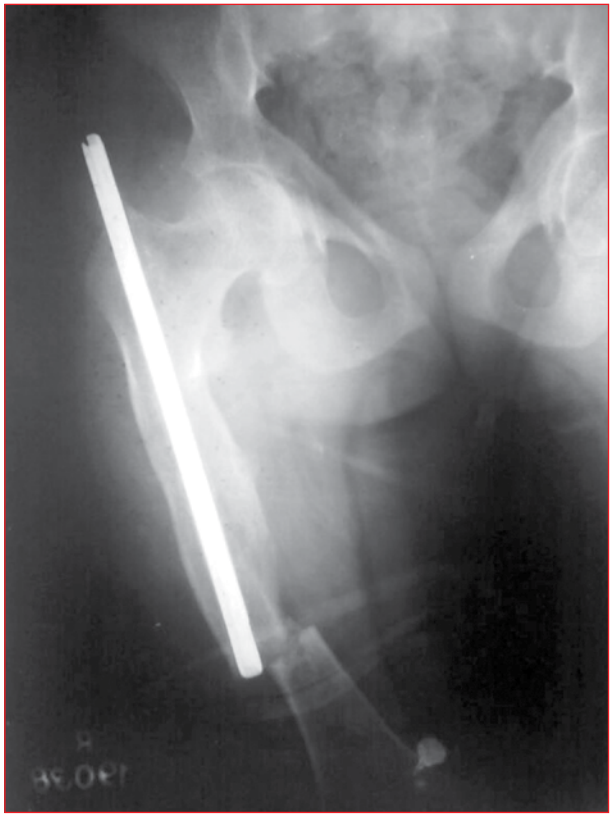

Figure 2: An X-ray showing a previous intramedullary nail in situ and the fractured femur

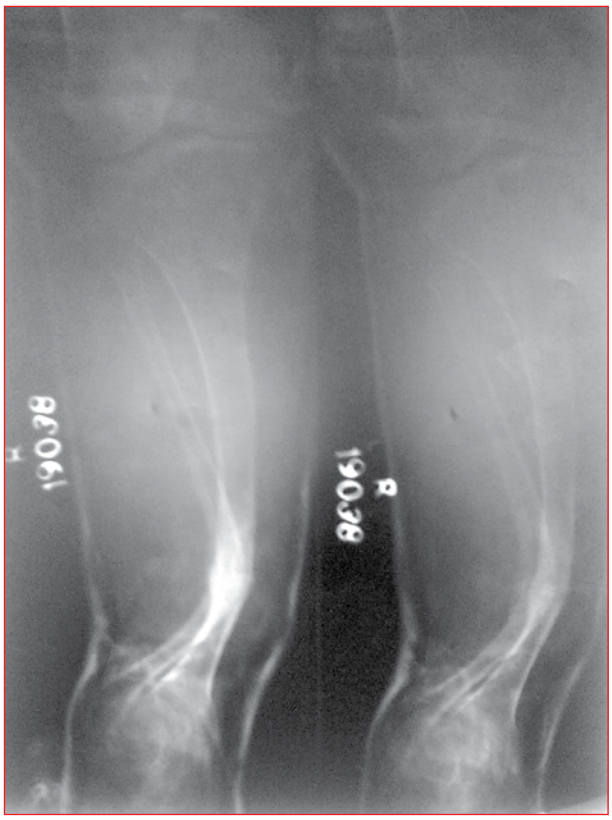

Figure 3: An X-ray showing valgus deformity of the leg

Table I: Investigations of the patient with osteogenesis imperfecta

Name of investigation	Result	Normal values
Haemoglobin	12 gm/dl	12-14 gm/dl
Bleeding time	2 minutes, 10 seconds	3-5 minutes
Clotting time	4 minutes, 15 seconds	4-11 minutes
Blood sugar	90 mg/dl	60-100 mg/dl
Blood urea	25 mg/dl	15-40 mg/dl
Serum electrolytes	*Sodium:* 135 meq/l	135-155 meq/l
	Potassium: 3.2 meq/l	3.5-5.5 meq/l
Total leukocyte count	9 000/mm^3	4 000-11 000/mm^3
Platelet count	250 000/mm^3	150 000-450 000/mm^3

The preoperative laboratory investigations were normal. The results are shown in Table I. The preoperative electrocardiography, echocardiography, chest X-ray and coagulation profile were also normal. A pulmonary function test was suggestive of mild restrictive disease. Airway examination revealed a mouth opening of 3 cm (two fingerbreadths), Mallampati class III score, with a short neck and defective dentition. Neck flexion and extension were limited. The patient was accepted for surgery as an American Society of Anesthesiologists (ASA) grade III patient. In view of the anticipated difficult airway and normal coagulation results, it was decided to proceed with a regional spinal anaesthetic technique. Before anaesthesia was conducted, a thorough preoperative examination and operating room preparation were completed, including the procurement of difficult airway equipment (a stylet, different sizes of face mask, different sizes of endotracheal tube, all sizes of laryngoscope blade, a laryngeal mask airway (LMA), a Proseal® LMA, a fibre-optic bronchoscope and a tracheostomy set).

The patient was positioned very carefully on the operation table to prevent a new fracture from occurring. An 18-G intravenous cannula was inserted on the dorsum of the right hand in the operation room. Standard ASA monitors, including electrocardiography, noninvasive blood pressure monitor and pulse-oximetry, were placed on the patient. The

baseline arterial pressure was 130/60 mmHg, the heart rate was 94 beats/minute, the respiratory rate was 18 breaths/minute, and the O_2 saturation on room air, 96%.

An intravenous preload of 500 ml Ringer's lactate was administered. Subarachnoid block was given in the sitting position in the space L3-4 with a 23-G Quincke spinal needle. Fentanyl (25 µg) was combined with 1.6 ml bupivacaine heavy 0.5% to achieve the desired block. The level of the block was up to T10. The patient was positioned cautiously. All of the pressure points were well padded. During surgery, her vitals remained stable. Rescue medication was not required throughout the intraoperative period. The operation lasted for one hour and 45 minutes. Estimated blood loss was 600 ml, which was adequately replaced. The patient's recovery was uneventful, as was her further stay in the hospital. She was discharged on the fifth postoperative day. Consent for her case to be reported was received from the patient.

Discussion

OI is a rare autosomal dominant inherited disease of connective tissue that affects bone, sclera and the inner ear.[8] The underlying cause is mutation in the gene coding for type 1 procollagen, i.e. collagen, type I, alpha 1 (COL1A1) and COL1A2. It affects 6-7 of 100 000 people, and occurs in approximately in 1:20 000 births.[5] Males and females are affected equally. No racial difference has been noted.[5] Initially, OI was divided into two forms: OI congenita and OI tarda. Fractures occur in utero, and death usually occurs in the perinatal period in the congenita form. Typically, the tarda form presents during childhood or early adolescence, and patients usually have a normal lifespan.[4] The most commonly used classification of OI by Sillence et al[9] categorises it according to four clinical types, I, II, III and IV, based on its phenotypic manifestations and the radiographic appearances of the bones. The latest classification divides OI into nine types.[6] The presentation of clinical severity depends upon the effect of mutation. Type I is considered to be the mildest form of the disease, and is compatible with long-term survival in adulthood. Types III-IX are moderate to severe forms of OI, depending on the defect in the structural proteins. Type II is lethal in the perinatal period and is not compatible with life.[6]

Anaesthetic management is influenced by coexisting orthopaedic deformities, vulnerability of fracture during simple positioning, platelet dysfunction, difficult intubation, cardiovascular abnormalities like mitral valve prolapse, a tendency to develop malignant hyperthermia, and rarely, extraskeletal manifestations.[10,11] Because of abnormal skeletal growth, odontoaxial dislocation and hypermobile joints, a difficult airway must always be anticipated in these patients.[12] Associated kyphoscoliosis with pectus carinatum may decrease vital capacity and chest wall compliance, with resulting arterial hypoxaemia due to ventilation perfusion mismatch.[2] This can pose an increased risk under general anaesthetic. Succinylcholine should be avoided because of its potential to cause malignant hyperthermia. Fasciculations can also lead to fractures.[13] Halothane is to be avoided as it may lead to malignant hyperthermia.[10,11]

Bergstorm, and Rampton, Kelly, Shanahan and Ingram, reported several cases of malignant hyperthermia in OI patients.[14,15] The general consensus is that many cases of hyperthermia in OI are not of the malignant type, but are the result of a hypermetabolic state instead. It has been suggested that hyperthermia is the result of either abnormal central nervous system temperature regulation or abnormal cellular energy metabolism.[13,16] At least 50% of patients with OI also have an elevated serum thyroxine level, leading to increased O_2 consumption and heat production.[5] Avoidance of common malignant hyperthermia-triggering drugs, temperature monitoring and the provision of necessary drugs and cooling devices should be easily available.[15] The fragility of bones in these patients is well known. Malde, Jagtap, Pantvaidya and Kenkare reported a fracture shaft of the femur in a patient with OI which occurred during the transfer of the patient to the recovery room.[12]

The best anaesthetic technique in these patients is regional anaesthesia as it avoids the necessity of tracheal intubation. The development of perioperative hyperthermia is also less likely to occur with regional anaesthesia than with general anaesthesia, and the former facilitates detection of a thyroid storm.[17] However, before administration of a regional anaesthetic, a coagulation profile must be undertaken because of the associated increase in bleeding time, despite the normal platelet count.[2] Chances of bone injury during positioning should be kept in mind.

Automated blood pressure cuffs may be hazardous as overinflation may result in a fracture.[17] Pressure points should be padded during prolonged surgery, and transportation must be gentle to prevent fracture occurrence.[12]

There have been several successful case reports of surgery being conducted under general, as well as regional anaesthesia on patients with OI.[1,2,12] Karabiyik, Parpucu and Kurtipek recommended total intravenous anaesthesia together with an intubating LMA to manage cases electively, while Malde, Jagtap, Pantvaidya and Kenkare successfully used balanced general anaesthesia in a case of OI with gross deformity of the pelvis for abdominal hysterectomy.[1,12]

We selected to use a regional anaesthetic technique since the patient had to undergo lower limb surgery and we wanted to avoid risks and complications relating to a general anaesthetic. Preanaesthetic investigations included routine investigations and a laboratory assessment of the

coagulation profile to eliminate a bleeding disorder, and a full cardiorespiratory work-up, including electrocardiography, echocardiography and pulmonary function tests, to discard associated abnormalities that are commonly associated with OI. General anaesthesia was avoided because of an anticipated difficult airway and associated restrictive lung disease, as well as susceptibility to malignant hyperthermia. A successful outcome was ensured by taking a careful history and an examination, and the application of basic principles in the management of the case.

To conclude, patients with OI pose a significant challenge to the anaesthesiologist owing to a difficult airway, problems with positioning, susceptibility to fractures, a tendency to develop perioperative hyperthermia, platelet functional abnormalities and difficult spinal anaesthesia. A detailed preoperative work-up, by taking a careful history and examination, as well as prompt management, can improve the outcome in these patients.

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