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Management of Anesthesia in Patient with Osteogenesis Imperfecta Type IV and Fractures on Tibia and Fibula: A Case Report

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ABSTRACT

Background: Osteogenesis imperfecta (OI), known as brittle bone disease, is a rare inherited and genetic skeletal disorder characterized by a significantly heightened risk of fragility fractures. About 1 in 15 to 20.000 births are affected with OI. these defects may moreover be combined with an airway that's troublesome to oversee, platelet dysfunctions, and other issues. The perioperative administration of anesthesia speaks to a significant challenge. Subsequently, it is critical to be able to anticipate the dangers during the perioperative period and to define and actualize particular high-quality anesthesia administration plans for such patients when they involve injury. **Case presentation:** We present a case of a female patient 26 years old with known OI type IV who underwent Osteotomy and Open Reduction Internal Fixation with plate and screw (ORIF PS) on her left Tibia and Fibula. In this case, we present the anesthesia technique with general anesthesia using a metal wire-enforced endotracheal tube to prevent partial obstruction due to kyphoscoliosis. **Conclusion:** It is crucial to assess the severity of the disease, identify risk factors in advance, and optimize the preoperative health status of individuals with OI.

1. Introduction

Osteogenesis imperfecta (OI), known as brittle bone disease, is a rare inherited and genetic skeletal disorder characterized by a significantly heightened risk of fragility fractures.¹ About 1 in 15 to 20.000 births are affected with OI, making it the most prevalent genetic cause of bone fragility.² The vertebrae and extremities are more susceptible to fractures due to the increased fragility of bone.³ The skeletal growth defect in Osteogenesis Imperfecta (OI) results from abnormal ossification of endochondral bone, leading to increased bone fragility, hypermobile limbs, and kyphoscoliosis.⁴ This disorder poses challenges for anesthesiologists due to various associated complications. These challenges include a

difficult airway (short neck, risk of odontaxial dislocation, difficult laryngoscopy, and intubation).³⁻⁵ Increased risk of fractures in cervical vertebrae, mandible, and teeth during these procedures. Other Associated Complications like Cardiac valvular lesions, neurologic abnormalities, metabolic abnormalities, Malignant hyperthermia, and platelet dysfunction can be found too. Managing anesthesia in individuals with OI requires careful consideration of these challenges. Anesthesiologists need to be aware of the potential complications and tailor their approach to ensure the safety and well-being of the patient.^{4,6} This may involve specialized techniques and equipment to address the unique characteristics of OI patients during surgical procedures.

2. Case Presentation

A 26 years old woman was known with OI Type IV since childhood. The patient came to the hospital diagnosed with pathological fracture on her middle third left Tibia and Fibula and planned for Osteotomy and ORIF PS surgeries. The injuries was obtained while she was sitting on the wheelchair, then the patient slipped and fell with her left leg hit the floor. The patient has complained pain on the injury site and felt more pain when moved or touched with numeric rating scale (NRS) up to 5/10. On Physical examination we found several notable findings. She had a short stature, measuring 105 cm, and weighed 35 kg. Vital signs indicated a blood pressure of 120/70 mmHg and a heart rate of 96 beats/min. The physical characteristics included fragile bones, and kyphoscoliosis. No signs of blue sclera, pallor, icterus, cyanosis, clubbing, lymphadenopathy, or edema were observed. The respiratory system examination no abnormalities with vesicular breath sounds and no additional lung sounds. The cardiovascular system examination showed normal heart sound, no murmur and gallop.

Additional tests, such as the electrocardiogram (ECG), liver function test, renal function test, and coagulation profile, all showed normal results. Chest X-ray showed no abnormalities in heart and lungs. There is thoracic levoscoliosis. Deformities found on first to eight ribs on the right in the posterolateral aspect and second rib on the left, suspected old fracture. Deformity accompanied by lytic lesions on third, fourth, fifth and sixth ribs on the left in the posterolateral aspect. We have consulted Pulmonologist to consider Spirometry examination, but the pulmonologist found no pulmonary problem so it was not necessary to do spirometry test. Airway assessment found acceptable flexion and extension at the neck, adequate mouth opening, and normal dentition, placing the patient in Mallampati class II. The patient was classified as American Society of Anesthesiologists Grade II with OI type IV and thoracic levoscoliosis as the actual problems. We decided to underwent anesthesia using general anesthesia with nonkinking (metal enforced) Endotracheal tube to avoid partial obstruction caused by the scoliosis.

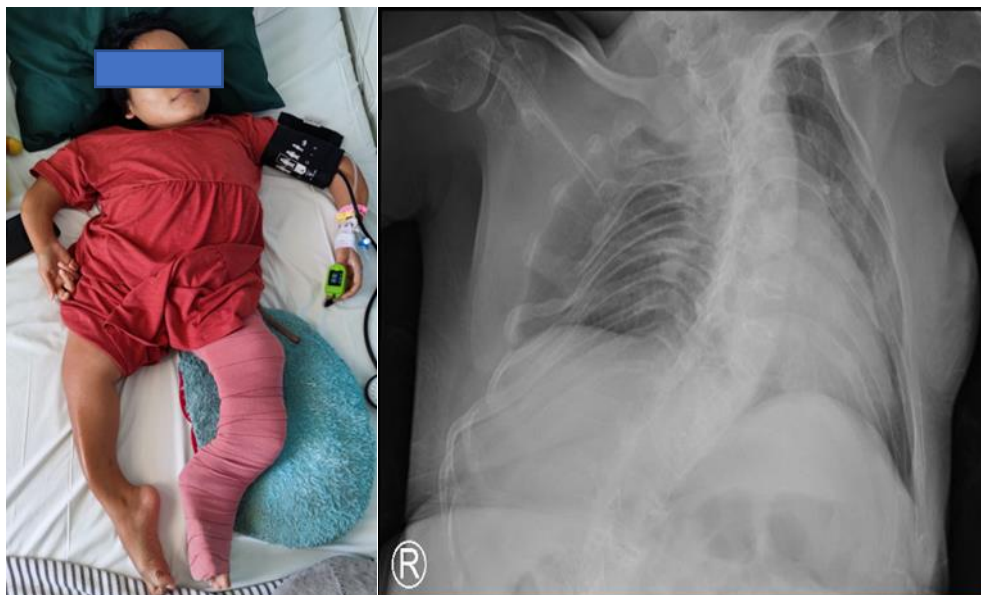


Figure 1. Clinical features and X-rays of patients.

During perioperative preparation, we make sure the difficult airway equipment is prepared. Intravenous access was checked too and we prepared

two IV lines because the difficulty to find IV line and the tendency of bleeding in this surgery. The patient was given 1 mg midazolam and Fentanyl 25 mcg

trough IV. After the pain decreased and the patient felt more calm The patient was positioned meticulously on the operating table, ensuring proper padding of pressure points. Monitoring equipment, including ECG, sphygmomanometer, and pulse oximetry (SpO₂), was applied to the patient to closely observe vital signs during the procedure. The patient was given Propofol as the sedation to maintained anesthesia using targeted controlled infusion (TCI), Schnider's mode with effect target 2-4 ng/mL We used intermittent Fentanyl for the analgesia during surgery. Muscle relaxation was used only for intubation with 15 mg of Atracurium. The surgeries took 65 minutes. The patient Vital signs was within normal limits, minimal bleeding and hemodynamically stable during the surgeries so we decided to extubated the patient. We found the ETT was completey 'S' shaped because of the scoliosis. The patient was observed in recovery room and transferred back to ward. The patient was given Morphine 20 mg for 24 hours IV and Paracetamol 500 mg every six hours orally for postoperative analgetics. The patient was discharged from hospital two days after the surgeries without experiencing any complications.

3. Discussion

OI is the most common inherited cause of bone fragility.⁶ The condition is most commonly caused by mutations in COL1A1 or COL1A2.⁴ These mutations result in the production of abnormal collagen (qualitative) or a reduced amount of collagen (quantitative), contributing to abnormal connective tissues and the characteristic features of OI. The most defining feature of OI is general osteopenia.^{1,4} The classification categorizes Osteogenesis Imperfecta into four types based on clinical and genetic features.⁷ The most common type is type I (quantitative), inherited in an autosomal dominant manner and is distinguished by dentinogenesis imperfecta, weak bones, hyperextensible joints, blue sclera, and gradual hearing loss. Type II (qualitative) is lethal during the perinatal or in utero stages. Both types III and IV have an autosomal dominant inheritance pattern. In type III

(qualitative), the patient typically passes away during childhood or adolescence due to heart problems making it the most severe type. Type IV (qualitative) is less severe than the other types, the patient has skeletal abnormalities but no abnormalities of the eyes, ears, or teeth.^{7,8} The majority of the literature on OI is made up of case reports; randomized controlled trials are minimal. Consequently, there is little information in the literature about the potential risks of Anesthesia using general, neuraxial and regional methods on patients with OI. For regional anesthesia with frequently encountered abnormal anatomy of extremities in patients with Osteogenesis Imperfecta (OI), obtaining peripheral nerve blocks may pose challenges. The use of nerve stimulation during these procedures could theoretically lead to contraction-induced fractures. Therefore, when administering peripheral nerve blocks in individuals with OI, ultrasound guidance is preferred over nerve stimulation to enhance safety.^{9,10}

A case series has reported successful anesthetic treatment of patients with OI using neuroaxial anesthesia procedures, including spinal anesthesia, epidural anesthesia, and caudal nerve blocks, but it is important to evaluate coagulopathy in OI. Due to short stature and anatomy variance, adjustments in epidural dosage should be made to ensure safety and efficacy. Individualized anesthesia management plans that consider the unique challenges associated with OI are crucial for optimizing patient outcomes. ^{9,10} In General anesthesia, the main problems are in the airway management and several reports of malignant Hyperthermia. One case arrangement portrays typical caffeine halothan contracture test (CHCT) comes about in patients with OI and detailed threatening hyperthermia (MH). In any case, there's one convincing report of a understanding with OI and MH who experienced common anesthesia. In rundown, prove of an affiliation between OI and MH is weak.^{5,9,11}

Managing the airway in patients with osteogenesis imperfecta (OI) poses unique challenges due to skeletal abnormalities, including thoracic kyphoscoliosis, decreased neck mobility, and the risk of atlanto-axial

dislocation or fractures with cervical spine overextension. Additionally, individuals with dentinogenesis imperfecta may be prone to tooth damage or loss during intubation. To address these challenges, the following considerations are crucial:

Anticipate difficult airway: Difficult airway management should always be assumed in patients with OI. The anesthesiologist must be prepared for potential difficulties in visualizing the larynx.⁵ Avoid cervical spine overextension to prevent atlanto-axial dislocation or fractures.⁵

Preoperative documentation: Patients with dentinogenesis imperfecta should have their dental abnormalities documented preoperatively to guide intubation strategies and prevent legal issues related to tooth damage.^{5,9,12}

Use of Video laryngoscopy: Video laryngoscopy is recommended as it reduces the need for head movement during endotracheal intubation, facilitating a safer procedure.^{5,9,12}

Incidence of airway difficulties: A case series of 205 anesthetic procedures in patients with OI reported an overall incidence of airway difficulties of 1.5%. The majority of difficulties occurred in patients with OI Type III. Videolaryngoscopic or fiberoptic endotracheal intubation successfully managed difficult airway situations.

Supraglottic Airway Devices: Laryngeal masks and other supraglottic airway devices have been used successfully and are considered essential, especially in emergency situations.

Individualized approach: An individualized approach to airway management, considering the specific skeletal and dental features of each patient, is crucial for a safe and successful procedure. In this case because of the kyphoscoliosis we used metal wired reinforced tube to avoid kinking and partial obstruction of the endo tracheal tube.^{5,9,12,13}

The use of suxamethonium (succinylcholine) in patients with Osteogenesis Imperfecta (OI) poses significant risks and should be avoided due to several reasons:

Lethal hyperkalemic responses: Patients with OI, especially those who are immobilized or denervated, may exhibit lethal hyperkalemic responses to suxamethonium injections. This is reported frequently and can result in serious

complications, including cardiac arrhythmias.

Up-regulation of acetylcholine receptors: The up-regulation of two new isoforms of acetylcholine receptors in immobilized or denervated body parts is thought to be responsible for the excessive release of potassium ions. This can lead to hyperkalemia, which is a life-threatening condition.^{4,9,11}

Immobilization of Patients: Patients with OI are often bound to wheelchairs or are otherwise immobilized, making them more susceptible to the adverse effects of suxamethonium.

contraction-induced fractures: There are reports in the literature documenting contraction-induced fractures after the administration of suxamethonium in patients with OI. This underscores the risk of using depolarizing neuromuscular blockers in this population.^{4,9,11} Given these risks, alternative strategies for muscle relaxation, such as non-depolarizing neuromuscular blockers, should be considered in patients with OI. Anesthesia providers should exercise extreme caution and tailor their approach to the specific needs and risks of each individual patient with OI.

In Osteogenesis Imperfecta (OI), there is evidence suggesting the presence of coagulopathy, which refers to abnormalities in the clotting or bleeding processes. Some observed coagulation-related changes in individuals with OI include platelet dysfunction, evidence indicates a decrease in platelet retention and reduced collagen-induced platelet aggregation. This suggests that the normal functioning of platelets, crucial for blood clotting, may be compromised in individuals with OI. Reduction in factor VIII Activity there was a reported reduction in factor VIII activity. Factor VIII is a clotting factor that plays a role in the blood coagulation cascade. A decrease in its activity may contribute to coagulation abnormalities. Capillary Fragility, coagulopathy in OI is associated with increased capillary fragility. This means that blood vessels, particularly small capillaries, may be more prone to rupture or bleeding. Considering these coagulation-related changes, it is recommended to complement routine platelet counts and standard coagulation tests with more specific assessments,

such as platelet function tests and measurements of factor VIII activity. This is particularly crucial when individuals with OI have a history of episodes of haemorrhagic diathesis (tendency to bleed excessively). It is important to be aware that individuals with more severe forms of OI may experience significantly higher intraoperative blood loss. This underscores the need for careful monitoring and management of coagulopathy during surgical procedures in individuals with OI.^{4,9,14}

Preoperative assessments and diagnostic tests are crucial for evaluating the overall health status of patients with osteogenesis imperfecta (OI) and planning appropriate anesthesia management. Here are some considerations. Spirometry tests, these tests help assess lung function and can reveal restrictive pulmonary disorders, particularly in patients with thoracic dysmorphologies. Blood Gas Analyses, preoperative blood gas analyses provide a baseline for gas exchange, facilitating later comparisons with intra- and postoperative values. Cervical spine X-ray, in patients with severe OI, there is a risk of basilar invagination and atlanto-occipital dislocation. X-rays of the cervical spine may be useful, especially if the surgical procedure involves complex patient positioning.^{4,9,10} Echocardiography, if the patient's medical history indicates symptoms of congenital heart defects or malformations of thoracic vessels, preoperative echocardiography can provide valuable information.^{9,10} These preoperative assessments help the anesthesia team tailor their approach to the specific needs and potential complications associated with OI. It allows for better planning and risk management during the perioperative period.

The use of automated blood pressure cuffs in patients with osteogenesis imperfecta (OI) is cautioned against due to the risk of over inflation, which could lead to fractures. To minimize this risk, healthcare providers are advised to use either invasive blood pressure monitoring or a manual sphygmomanometer.^{9,15} Furthermore, when caring for patients with OI, special attention should be given to padding pressure points adequately to prevent injury.

A thorough preoperative workup is essential, including an assessment of the patient's coagulation profile and a comprehensive cardiorespiratory evaluation. This evaluation may involve electrocardiography (ECG), echocardiography, and pulmonary function tests to ensure a comprehensive understanding of the patient's cardiovascular and respiratory status.^{9,15} These precautions and thorough assessments contribute to the development of a safe and tailored anesthesia management plan for individuals with OI, considering the unique challenges associated with this condition.

4. Conclusion

Osteogenesis imperfecta (OI) is a serious disease with multiple complications, emphasizing the importance of a comprehensive preoperative evaluation and the development of an individualized anesthesia management plan for affected patients requiring surgery. To ensure safety and smooth anesthesia, it is crucial to assess the severity of the disease, identify risk factors in advance, and optimize the preoperative health status of individuals with OI. The recognition of specific challenges associated with OI, such as brittle bones, joint hypermobility, and the risk of fractures during intubation, allows for the implementation of tailored strategies to mitigate these risks. Anesthesia management plans should consider the unique characteristics and potential complications associated with OI, and healthcare providers should collaborate closely to share insights and experiences in managing these cases. The overall goal is to provide safe and effective anesthesia while minimizing the potential risks and complications associated with the surgical and perioperative care of individuals with OI.

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