Clinical Case Reports International

Case Report
Published: 23 Sep, 2022

Lifelong Complications of Osteogenesis Imperfecta: Case Report and Literature Review

Liviu-Iulian L, Ungureanu R, Toma M, Angheluță C, Ioana-Marina G and Mirea LE*

Department of Anesthesia and Intensive Care, Clinical Emergency Hospital of Bucharest, Romania

Abstract

Osteogenesis Imperfecta (OI) is a rare inherited connective tissue disorder with many phenotypic presentations, often called "brittle bone disease". Diagnosis is straight forward in individuals with bone fragility and a positive family history or several extra skeletal manifestations (hearing loss, dark or bluish sclera). OI is classified into a number of major subtypes based on genetic, radiographic, and clinical characteristics, but more useful is the clinical classification, based upon the typical problems that manifest in infants, children, and adults with mild, moderate to severe and lethal disease.

This case report describes a 53-year old female patient with an unknown genetic disorder, but with medical history of bone fragility, currently presented with spontaneous ace tabular fracture that led to the diagnosis of osteogenesis imperfect a type I. OI is associated with multiple perioperative complications, thus the patients require extensive preoperative evaluation and an individualized surgical and anesthetic management.

Keywords: Osteogenesis imperfecta; COVID-19; Periarticular

Introduction

Osteogenesis imperfecta is rare inherited connective tissue disorder, being characterized by bone brittleness and increased fracture risk. Its incidence is described at 1:10.000-20.000 live births [1]. Clinical diagnosis includes fractures from the mildest of trauma, short stature, bone deformities, blue sclera, dentinogenesis imperfecta and deafness [2]. Clinical severity varies from mild forms with only premature osteoporosis or severe postmenopausal bone mineral loss determining normal life expectancy in OI type I, to perinatal death in OI type II. Almost 90% of cases are caused by mutations in the genes encoding type I collagen [3]. Perioperative management can be challenging due to difficult airway, respiratory failure due to scoliosis or abnormal platelet function with easy bruisability. It is essential to predict these risks and implement a high-quality management for trauma in such patients. Treatment of OI depends on the severity and age, the main objectives being to maintain autonomy and prevent long term complications [4]. This current case report describes a patient with an unknown genetic disorder and her medical history that led to the diagnosis of osteogenesis imperfecta.

Materials and Methods

In September 2021, a 53 year-old female (body mass 78 kg, height 157 cm, body mass index 31.6 kg/m^2) was admitted for a minor trauma (same height fall), which resulted in a complex left acetabular fracture and protrusio acetabuli (Figure 1).

Clinical examination revealed blue sclera (Figure 2), moderate scoliosis, and mild skeletal deformities of the forearms, dentinogenesis imperfecta and deafness.

Twenty years prior she developed early onset deafness and underwent a stapedectomy for presumably right ear otosclerosis. Her hearing worsen in both ears over the next six years and needed hearing aid. That's when she was first told she had a genetic disorder, but at the present time she did not possess any medical history, nor remembered any details. Family history revealed the patient's mother also had blue-gray colored sclera. The patient had multiple rheumatology admissions for osteoporosis, tendinitis and bilateral chronic hip and knee articular pain requiring treatment with analgesics and bisphosphonates (alendronic acid). She also had a history of essential arterial hypertension, gastro esophageal reflux disease and smoking. She required immediate plate osteosynthesis of the posterior column of acetabulum (Figure 3) with normal perioperative evolution, being discharged in good clinical condition.

OPEN ACCESS

Correspondence:
Liviu-Iulian Lovin, Department of Anesthesia and Intensive Care, Clinical Emergency Hospital of Bucharest, Calea Floreasca, Nr. 8, Sector 1, Bucharest, Romania,
E-mail: spital@urgentafloreasca.ro

Received Date: *06 Sep 2022*
Accepted Date: *20 Sep 2022*
Published Date: *23 Sep 2022*

Citation:
Liviu-Iulian L, Ungureanu R, Toma M, Angheluță C, Ioana-Marina G, Mirea LE. Lifelong Complications of Osteogenesis Imperfecta: Case Report and Literature Review. Clin Case Rep Int. 2022; 6: 1403.

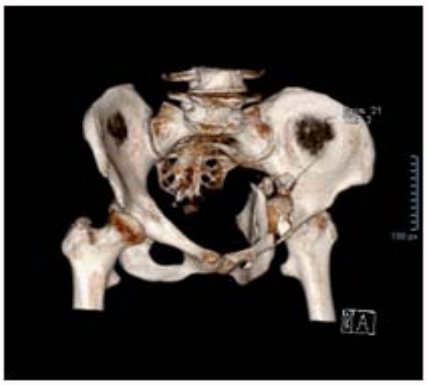

Figure 1: Left acetabular fracture and protrusio acetabuli.

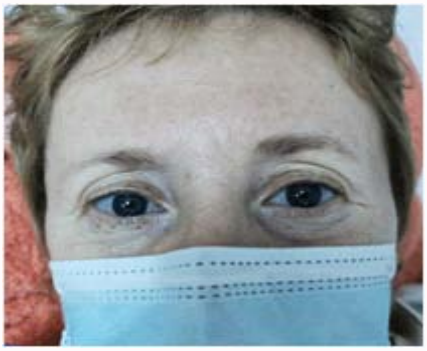

Figure 2: Blue sclera.

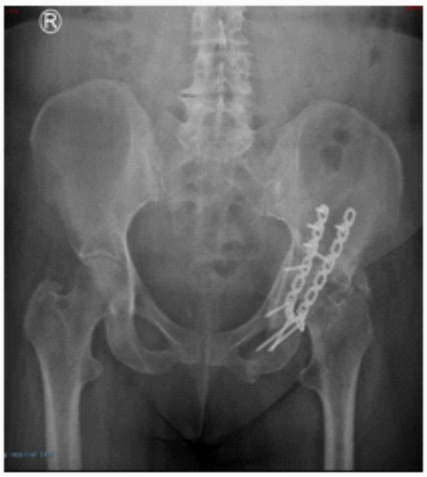

Figure 3: Ostheosynthesis of the posterior column of acetabulum.

During recovery, in November 2021 she develops respiratory signs of SARS-CoV-2 infection (fever, cough, muscle aches and sore throat) and is diagnosed with a positive RT-PCR test through her family doctor. She had a mild to moderate form and received inpatient low-flow supplemental oxygen therapy *via* nasal cannula and 8 mg daily dexamethasone for 5 days.

The approach to management of COVID-19 evolves rapidly as clinical data emerge, but corticosteroids are almost a constant in treatment protocols for moderate to severe cases [5]. The use of high doses of dexamethasone was lifesaving in many cases, but one adverse effect is corticosteroid induced a vascular necrosis of the femoral head

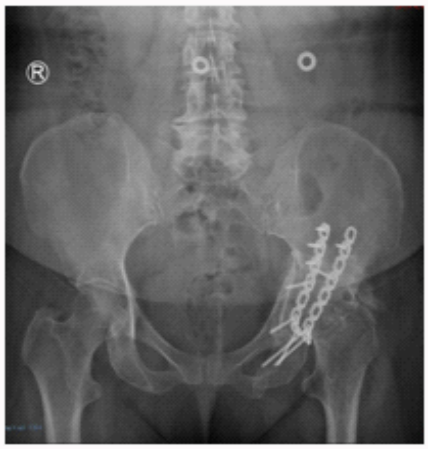

Figure 4: Aseptic necrosis of the left femoral head.

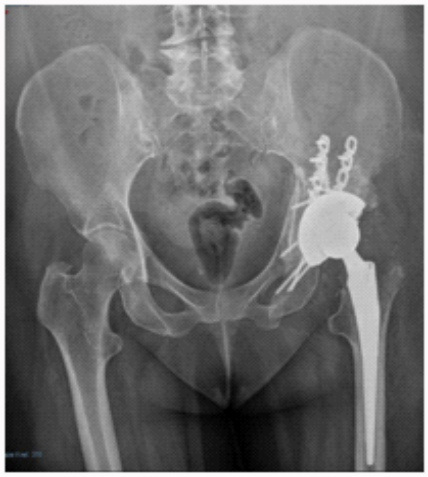

Figure 5: Total left hip arthroplasty.

[6]. Corticosteroids induce fat mobilization and may promote fat emboli occlusion of blood vessels, disrupt calcium metabolism and increase osteoclasts activity and bone resorption [7]. In January 2022, on a follow up visit the patient had recurrence of acute, severe, left hip articular pain. Although all her previous follow-up visits shown progressive healing of the fracture, this time her radiology report showed aseptic necrosis of the left femoral head (Figure 4), requiring total hip arthroplasty (Figure 5).

The patient received general anesthesia combined with an ultrasound-guided fascia iliaca block to ensure adequate perioperative analgesia, with good outcome. General anesthesia was preferred over neuroaxial anesthetic procedures due to the coagulopathy found in OI patients which includes reduced platelet aggregation, reduced factor VIII activity and an increased capillary fragility. Difficult airway must be anticipated in these patients and video laryngoscopic or fiber optic endotracheal intubation should be readily available. The moderate scoliosis with restrictive lung disease and long-term effects of COVID-19 like lung fibrosis can impede perioperative respiratory function.

Results and Discussion

A vascular necrosis of the femoral head can be a consequence of the trauma, it can be corticosteroid-induced, but OI can also

associate periarticular bone dysplasia and lesions of ischemic origin. Taking that into account, the differential diagnosis can prove difficult in a patient with history of osteoporosis, chronic hip pain and corticosteroid treatment. Considering all of the above, a clinical diagnosis of osteogenesis imperfecta type I was formulated.

OI is also known as "brittle bone disease" because of high risk of fractures after mild trauma, or ever from transporting or mobilization of patients. Diagnosis of OI on the basis of clinical and radiological findings is straightforward in most cases, but bone mineral density measurements, bone metabolic markers and morphological bone studies may aid the process. Genetic testing can be useful in differentiating between quantitative and qualitative collagen I defects. Bone fragility, osteoporosis and coagulopathy can complicate or prolong surgical interventions. OI is associated with multiple complications, thus the patients require extensive preoperative evaluation and an individualized orthopedic and anesthetic management.

Conclusion

Currently, only symptomatic therapy is available, such as growth hormone or bisphosphonates and surgical treatment is necessary for decreasing the rate of complication. Most of the literature focuses on diagnosis and management of OI in the pediatric population, but the manifestations are lifelong and some cases can be first diagnosed throughout adulthood. In this case the new onset of aseptic necrosis of the femoral head was pluri factorial, corticotherapy-induced, a complication of bone brittleness and ischemic lesions found in OI, as well as the recent history of acetabular fracture. The approach to such a heterogeneous disease should be multidisciplinary in order to assure adequate long-term quality of life and decreasing complication rates.

References

1. Marini JC, Forlino A, Bachinger HP, Bishop NJ, Byers PH, Paepe A, et al. Osteogenesis imperfecta. Nat Rev Dis Primers. 2017;3:17052.
2. Oakley I, Reece LP. Anesthetic implications for the patient with osteogenesis imperfecta. AANA J. 2010;78(1):47-53.
3. Marini JC, Forlino A, Cabral WA, Barnes AM, San Antonio JD, Milgrom S, et al. Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: Regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. Hum Mutat. 2007;28(3):209-21.
4. Antoniazzi F, Mottes M, Fraschini P, Brunelli PC, Tato L. Osteogenesis imperfecta: Practical treatment guidelines. Paediatr Drugs. 2000;2(6):465-88.
5. Ye Z, Wang Y, Colunga-Lozano LE, Prasad M, Tangamornsuksan W, Rochwerg B, et al. Efficacy and safety of corticosteroids in COVID-19 based on evidence for COVID-19, other coronavirus infections, influenza, community-acquired pneumonia and acute respiratory distress syndrome: A systematic review and meta-analysis. CMAJ. 2020;192(27):E756-67.
6. Zhang S, Wang C, Shi L, Xue Q. Beware of steroid-induced avascular necrosis of the femoral head in the treatment of COVID-19 experience and lessons from the SARS epidemic. Drug Des Devel Ther. 2021;15:983-95.
7. Xie XH, Wang XL, Yang HL, Zhao DW, Qin L. Steroid-associated osteonecrosis: Epidemiology, pathophysiology, animal model, prevention, and potential treatments (an overview). J Orthop Translat. 2015;3(2):58-70.