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Complications among patients with osteogenesis imperfecta following surgical interventions

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ABSTRACT

Objectives: Osteogenesis imperfecta (OI) is a genetic bone disorder caused mainly by a defect in the COL1A1 or COL1A2 genes. This study aimed to analyze the complication using surgical interventions, including intramedullary telescopic rod fixation, Rush pin fixation, and Kirschner-wire (K-wire) fixation.

Methods: This retrospective and cohort study was conducted in our institution, Riyadh, Saudi Arabia. A total of 23 patients with OI were treated from 2018 to 2022, out of which 19 were treated surgically. A total of 29 long bones were surgically managed. Our patients' data were collected from the hospital's electronic system.

Results: The overall incidence of Rush pin fixation post-operative complications was 57% (five out of seven cases) and K-wire fixation was 50% (one out of two cases). The main indications for intramedullary telescopic rod implantation were osseous deformities and fractures, with the overall incidence of post-operative complications reaching around 35% (seven out of 20 cases).

Conclusion: In our population, the overall incidence of complications after intramedullary telescopic rod fixation was lower than those after Rush pin fixation and K-wire fixation. In addition, more studies are needed to add to the literature new ways of tackling post-operative complications following the insertion of intramedullary telescopic rods, such as nail migration and new fractures or refractures.

Keywords: Complications, Intramedullary telescopic rod, Morbidity, Osteogenesis imperfecta, Surgical interventions

INTRODUCTION

Osteogenesis imperfecta (OI) is a genetic bone disorder caused mainly by a defect in the COL1A1 or COL1A2 genes, making bones vulnerable to fractures and deformities. It leads to an abnormal cross-linking process and an overall decrease in Type 1 collagen.[1] The prevalence of OI is estimated to be approximately one in 15,000–20,000 births.[2] This bone disorder has orthopedic manifestations such as bone fragility and fractures, ligamentous laxity, short stature, scoliosis, basilar invagination, radial head dislocations, bone deformities, and many other manifestations.[3] Up to this moment, OI diagnosis is still based mainly on family history associated with typical radiographic and clinical features, as no diagnostic tests are available due to the wide variety of genetic mutations that can cause OI.

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OI is a highly complex disorder, and its classification and definition can be challenging due to its diverse manifestations. The severity of OI varies widely, with some forms being lethal in the perinatal period, while others may be mild and difficult to identify as patients may appear to be near normal individuals. Clinical classifications can be useful in certain situations to determine the severity of the disorder. Shapiro's classification[4] and Sillence's classification[5] are two such methods that rely on clinical criteria. Initially, Sillence's classification involved four types, then Types V and VI were added by Glorieux[6,7] and later, Type VII was added by Ward [Table 1].[8] Although some of these classifications are being used today, the author of the most commonly used classifications, Sillence, believed that more than 12 types of OI exist.[9]

The most effective management strategy for OI involves a multidisciplinary approach that prioritizes fracture prevention through medications such as bisphosphonates. In addition, the approach involves managing fractures using cast immobilization and providing surgical treatment when fractures are present or realignment osteotomies to correct long bone deformities when necessary.

Historically, patients with OI who were suffering from fractures or deformities that indicate surgical interventions have been treated using Rush pin fixation. Later, new surgical techniques like intramedullary telescopic rods emerged, becoming the gold standard surgical treatment to fix and even prevent long bone fractures. The moderate forms of OI can be treated using this option in an almost minimally invasive manner. However, severe cases of OI are still challenging to treat and impose new complex surgical techniques. Regardless of the surgical option used to treat patients with OI, the outcome, complications, and morbidity rates following surgery compared to conservative treatments are still not reported in our region, and the studies done about this topic are few.

A study from Riyadh, Saudi Arabia, evaluated the ambulatory status in patients with OI before and after the lower limb Sofield procedure.[10] In that study, the ambulatory status after the Sofield procedure was assessed using Hoffer and Bullock's grading system. Only 3% of the patients improved and around 54.6% worsened after surgery. On the other hand, another study by the same authors reported improved upper limb function in all patients after the Sofield procedure.[11] The difference in outcomes shown by these two studies depending on the upper versus lower limb being affected can help surgeons in the decision-making process when choosing the Sofield procedure, as the outcomes favor performing the Sofield procedure to the upper limbs but not to the lower limbs.

Some studies, however, have reported a high incidence of complications following surgical treatment of OI. For instance, a study published by Jerosch *et al*. reported a high total complication rate reaching 63.5%.[12] Furthermore, a study from Oman reported a complication rate of around 13.5% in OI patients after a modified Sofield procedure.[13]

Although a few regional studies discussed the complication rates after some surgical procedures in patients with OI, no studies in the region discussed the complication rates after intramedullary telescopic rod fixation or Rush pin fixation in patients with OI. Therefore, this study aimed to analyze and identify the complication rates among patients with OI who underwent surgical interventions, including intramedullary telescopic rod fixation, Rush pin fixation, and Kirschner-wire (K-wire) fixation in a tertiary hospital in Riyadh, Saudi Arabia.

Table 1: Sillence's classification, modified by Glorieux and Ward.

Type	Genetics	Severity	Teeth	Sclerae	Other characteristics
I	Autosomal dominant	Light form	Normal	Blue	
II	Autosomal recessive	Lethal form	Dentinogenesis imperfecta	Blue	
III	Autosomal recessive/ dominant	Severe form	Dentinogenesis imperfecta	Bluish	
IV	Autosomal dominant	Moderate form	Dentinogenesis imperfecta or normal	White	
V	Autosomal dominant	Moderate form	Normal	White	Hypertrophic calluses, limited pronation, and supination of the forearms (calcification of the interosseous membrane)
VI	Probably autosomal dominant	Extremely rare moderate form	Dentinogenesis imperfecta or normal	White	Vertebral fractures, elevated alkaline phosphatase
VII	Autosomal recessive	From moderate to extreme	Dentinogenesis imperfecta or normal	White/Blue	Rhizomelic form (disharmonic pelvic and thoracic limbs due to the involvement of the femur and humerus, Dwarfism, and Coxa vara

MATERIALS AND METHODS

This retrospective and cohort study was conducted in King Fahad Medical City (KFMC), Riyadh, Saudi Arabia. A total of 23 patients with OI were treated from 2018 to 2022 in KFMC. All patients diagnosed with OI who underwent surgical intervention from 2018 to 2022 were selected. Surgical treatment was the option used in 19 patients. Four patients with a mild type of OI were conservatively managed by physical therapy with frequent follow-ups, and they were excluded from our study. The surgical interventions included Rush pin fixation, intramedullary telescopic rods, and K-wire fixation. Our patients' data were collected from the hospital's electronic system as this was a chart review study. Numerical data in this study were presented using means and standard deviations.

RESULTS

At the initial surgical procedure, the average age of our patients was 8.2 years. The average follow-up was 1.4 years. During the 5 years between 2018 and 2022, 29 long bones were surgically managed. Table 2 illustrates the distribution of operations between the upper and lower limbs. Seven long bones were managed using Rush pin fixation and two long bones were managed using K-wires because the intramedullary canal diameter was smaller than the diameter of the smallest available rod. The remaining 20 long bones were stabilized using intramedullary telescopic rods.

The overall incidence of Rush pin fixation post-operative complications in our population was around 57% (five out of seven cases). Three cases of femur nail migration and one case of tibial nail migration with perforation of the knee joint are the rush pin fixation cases with post-operative complications [Figure 1]. Similarly, the incidence of post-operative complications of K-wire fixation was 50% (one out of two cases). One case had surgical site infection and soft-tissue perforation after K-wire fixation of the right ulna. The main indications for intramedullary telescopic rod insertion were bone deformities (12 long bones, 60%) and fractures (8 long bones, 40%) [Figure 2]. The intraoperative complications with the distribution of complications among the long bones after intramedullary telescopic rod fixation are shown in [Table 3]. The complications that were found directly after surgery in six patients were revised as soon as they were noted, and patients were taken back to operating rooms right away to correct the position and/or length of the implants used. The overall incidence of post-operative complications was around 35%

Table 2: Distribution of operations between the upper and lower limbs.

Extremity	Number of intramedullary telescopic rod fixations	Number of Rush pin fixations	Number of Kirschner wire fixations
Femur	12	5	0
Tibia	3	2	0
Fibula	1	0	0
Humerus	2	0	1
Ulna	2	0	1

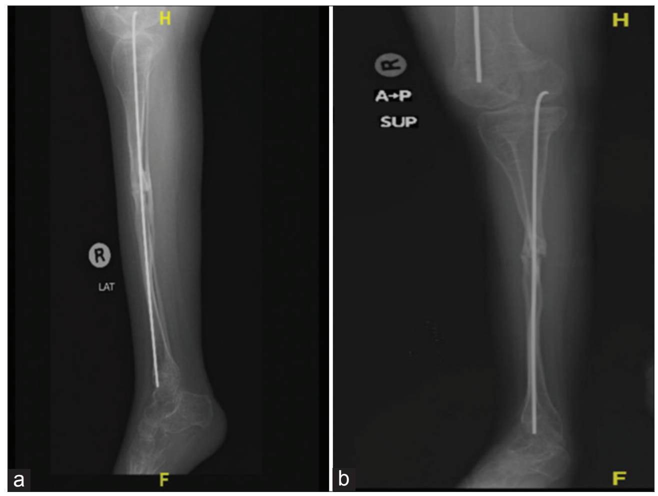

Figure 1: (a and b) Anteroposterior and lateral views of a 15-year-old female who underwent Rush pin fixation for right tibia deformity showed superior Rush pin migration causing the right knee joint perforation.

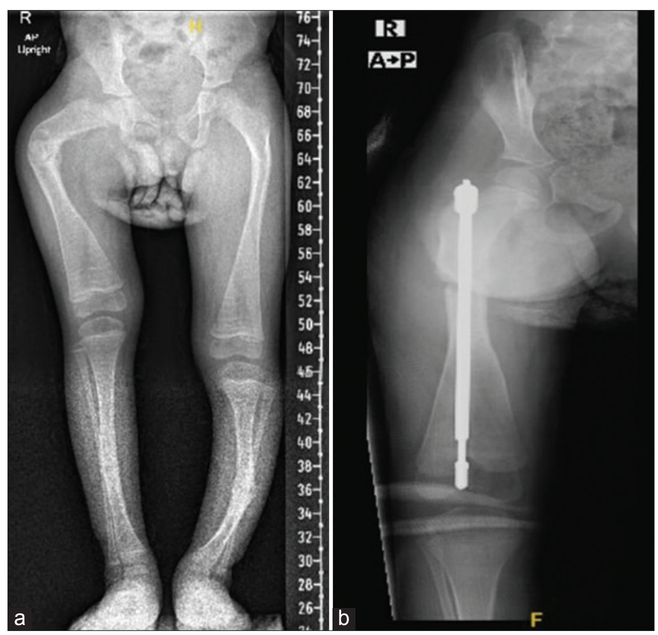

Figure 2: (a and b) Radiographs of a 3-year-old female patient with the right femur deformity before and after surgery. The radiograph taken after surgery shows improvement of the deformity using the intramedullary telescopic rod fixation.

Table 3: Intraoperative complications of intramedullary telescopic rod fixation.

Extremity	Number (*n*)	Complications			
		Nail too long proximally	Nail too short proximally	Nail too long distally	Nail too short distally
Femur	2	1	0	1	0
Tibia	2	0	1	1	0
Humerus	1	0	0	1	0
Ulna	1	1	0	0	0
Total	6	2	1	2	1

Table 4: Distribution of postoperative complications of intramedullary telescopic rod fixation.

Complication	Total	Percentage
Nail migration	4	20
New fractures or refractures	3	15
Deformity recurrence	2	10
Infections	2	10
Bone perforation	1	5
Lack of elongation	1	5
Nail exostosis	1	5
Persistent deformity	1	5
Non-union	1	5
Soft-tissue perforation	0	0
Bending or fracture of the nail	0	0
Pseudoarthrosis	0	0

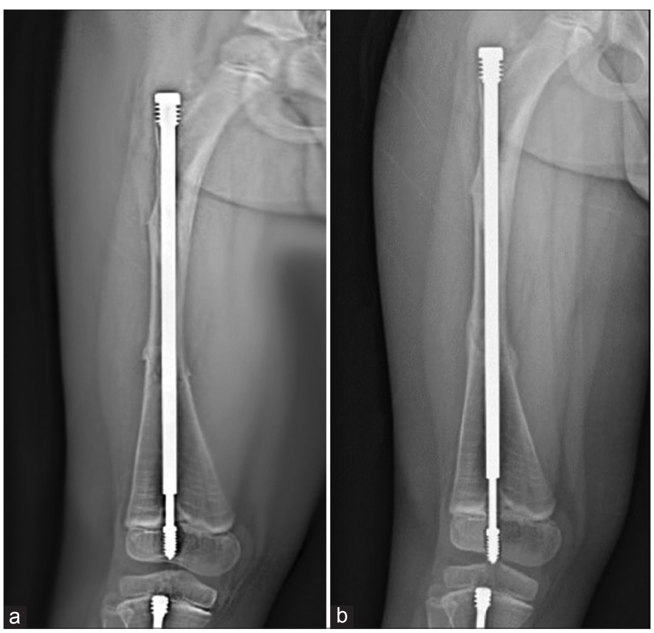

Figure 3: (a and b) Anteroposterior view radiograph of a 2-year-old male patient with the right femur deformity after double osteotomy and intramedullary telescopic rod insertion. The radiograph on the left side was taken immediately after surgery, and the one on the right side was taken during follow-up and showed the right knee joint perforation by the male part of the femoral intramedullary telescopic rod.

(seven cases out of 20) [Table 4]. Most patients who suffered from post-operative complications had more than one complication. After intramedullary telescopic rod implantation, nail migration [Figure 3] showed the highest incidence (four operated bones) and migration happened despite using some well-known techniques to avoid this complication like centralizing the insertion point of the male component into the epiphysis as well as ensuring sufficient depth of purchase. The second most common complication was new fractures (three operated bones).

One bone has shown persistent deformity postoperatively. In two cases, there was a recurrence of deformity after intramedullary telescopic rod insertion. None of our patients has developed pseudarthrosis after surgery. The telescoping rod did not extend in one case, but no rod was disassembled or separated. Bone perforation by the intramedullary telescopic rod happened in one case, but it did not lead to the perforation of the soft tissue around the rod. Knee joint perforation by the intramedullary telescopic rod happened in two cases after the rod insertion. One intramedullary telescopic rod that was inserted in a femur migrated proximally into the abductor's muscles. We also found in one case that the rod perforated the ankle joint distally. Two surgical site infections were encountered in one rod applied to a femur and another to a humerus. In one case, a humeral bone non-union occurred due to the failure of immobilization to the fractured bone pieces by the cast that was applied to augment the surgical fixation.

DISCUSSION

Few regional studies about the complications of intramedullary telescopic rods for the treatment of OI have been published in the past 10 years. The number of studies in Saudi Arabia is still low, perhaps due to the fact that OI is a rare disease and because the management of OI is still challenging for many orthopedic surgeons and health-care systems. While many hospitals and centers worldwide lack the infrastructure, the proper advanced equipment, and the expertise in this technically demanding condition, the number of research papers about the surgical management of this condition is expected to be low. Nevertheless, one of the published studies reported that the incidence rate of complications post-intramedullary telescopic rod implantation in patients with OI was around 63.5%.[12] This is relatively higher than our reported incidence rate of complications; however, our sample size is smaller compared to that study. Moreover, the implants used in that study were different from those used in ours. Furthermore, the improved level of training and the new and better equipment versions might have contributed to the lower rates of complications nowadays compared to the time of that study around three decades ago.

In our study, the most frequently reported complication was nail dislocation or migration into joints. In most published studies, this complication is almost always the most common. One of the logical explanations for that is because of repetitive load over weak cancellous epiphyseal bones in skeletally immature patients suffering from OI. The most common joint affected among our patients was the knee joint, followed by the ankle. With new equipment versions, separation or disassembly of the fixation device was not encountered. During follow-up of our patients, we also noticed some patients' related factors that can lead to higher rates of complications postoperatively, such as poor compliance with medications, poor weight control, many losses of follow-ups or delayed follow-ups, little or no physical therapy after surgery, and most of the time it is due to family neglect.

Some intramedullary telescopic rods have been developed to have lower complication rates. For instance, the Fassier-Duval intramedullary telescopic rod has advantages over other intramedullary telescopic rods, such as fewer revision procedures, improved walking, and better survival rates.[14,15] Although intramedullary telescopic rod fixation is currently the surgical standard of care for patients with OI, it has some drawbacks, such as the high cost, the long learning curve to master the surgical steps and post-operative complications.

One of the main limitations of our study was the small sample size; thus, our results cannot be generalized to a large population. This was due to the fact that OI is a relatively rare disease in our community, the indications for surgery are few, and the high number of patients who, at some point, stopped showing up in pediatric orthopedic clinics. Another limitation of our study was the different surgeons operating in our team. As we had different surgeons from different backgrounds with different skills and experience levels managing our patients, we had to consider that when analyzing the complication rates after each surgical option. Furthermore, some of the complications, such as bleeding and blood transfusion complications, were not collected in this study, unfortunately, as there were some missing documents from some of the patients' files after surgery.

CONCLUSION

The overall rate of complications after intramedullary telescopic rod fixation was lower than the incidence rate after Rush pin fixation and K-wire fixation in our population. This supports the current standard of choosing intramedullary telescopic rod fixation for fractures or deformities in patients with OI. In addition, more studies are needed to add to the literature new ways of tackling post-operative complications following the insertion of intramedullary telescopic rods, such as nail migration and new or refractures.

Recommendations

Orthopedic surgeons must be familiar with the common complications and how to prevent them as much as possible, and the patient and the patient's family must be educated about the high risk for complications and the possibility of revision procedures. More researchers are needed to contribute to the literature about this condition and how we can find new solutions to prevent or lower the currently high complication rates.

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AUTHORS' CONTRIBUTIONS

MMA conceived and developed the idea of the study, carried out the proposal formation, and helped in the data analysis phase. AF contributed to the planning and execution of this study and supervised the findings of this work. RH contributed to the design and analysis of the results and the writing of the manuscript. AA contributed to data collection, manuscript writing, and editing. All authors have critically reviewed and approved the final draft and are responsible for the manuscript's content and similarity index.

USE OF ARTIFICIAL INTELLIGENCE (AI)-ASSISTED TECHNOLOGY FOR MANUSCRIPT PREPARATION

The authors confirm that there was no use of Artificial Intelligence (AI)-Assisted Technology for assisting in the writing or editing of the manuscript and no images were manipulated using the AI.

ETHICAL APPROVAL

Institutional Review Board, King Fahad Medical City, issued approval on December 27, 2022, IRB Log Number: 22-617.

DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patients consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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CONFLICTS OF INTEREST

There are no conflicting relationships or activities.

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