Original Article

Results of Rodding and Impact on Ambulation and Refracture in Osteogenesis Imperfecta: Study of 21 Children

Atul R Bhaskar, Deepak Khurana[1]

Consultant Paediatric Orthopaedic Surgeon and [1]Clinical Fellow – Paediatric Orthopaedics, Children Orthopaedic Surgical Services, Mumbai, Maharashtra, India

Abstract

Introduction: Delay in presentation and surgical intervention is quite usual in osteogenesis imperfecta (OI) because of various local and cultural beliefs. The purpose of this study is to review the results of 21 children who had intramedullary rodding and its effect on ambulation and refracture. **Methods:** We reviewed 21 children with a clinical diagnosis of OI. The mean age of children at presentation was 8.74 years (3–21 years). All children had recurrent fractures of long bones. Twenty eight femurs and 21 tibiae were stabilized with intramedullary rodding. Ambulatory status was assessed by the Hoffers and Bullock's (H and B) grading, and muscle power was recorded using the Medical Research Council, U. K., grade. Ten children had received intravenous bisphosphonates preoperatively. Postoperatively, the children were assessed for ambulatory status, pain, and ability for independent self-care. **Results:** The mean followup period was 34 months (24–48 months). Rush rods were used in 20 femurs, the Fassier–Duval (FD) rods in 6 femurs, and in two cases, with narrow intramedullary canals, Kirshner (K) wires were used. For the tibiae, 15 children received rush rods and in 6 cases, an FD rod was used. The mean time to fracture union was 8 weeks (6–12 weeks). Before surgery, 13 children were in H and B Grade 4 (wheel-chair independent or carried by parents usually in a developing country), four were able to ambulate with a walking aid (H and B Grade 3b), and four children were able to walk about in the house without aids (H & B Grade 2). After the rodding procedure, the ambulatoty status improved in 11 (50%) children. Seven children (33%) became household physiologic walkers (H & B Grade 3b), three achieved independent ambulation with orthosis (H & B Grade 1b), and one child with mild OI could walk unaided (H & B Grade 1a). No child had deterioration in ambulatory status. Only two children had refractures at the distal end of the rod due to continual growth of bones. **Conclusions:** Intramedullary rodding treatment for recurrent fractures in children with OI improves their mobility potential. It also and prevents repeated cast application, disuse wasting, and osteopenia which can lead to deterioration in the quality of the long bones.

Keywords: *Ambulation and rodding, refracture, osteogenesis imperfecta*

Introduction

Osteogenesis imperfecta (OI) is a genetically heritable disorder of Type 1 collagen synthesis, either qualitative or quantitative.[1,2] Clinically, it is characterized by low bone mass, increased bone fragility, recurrent fractures, and multiapical deformities in long bones.[3] The most common defect is caused by multiple mutations in COLA1 and COLA2 genes located on chromosome 17 and 7, respectively.[4] The defective collagen leads to wide spectrum of phenotypic presentations in OI that range from milder forms with no fractures to forms that are lethal in the perinatal period.[5] Based on genetic and clinical features, Sillence *et al.* classified OI into four major types and many other sub types.[6-10] Type 1 OI has milder disease, Type 2 is lethal at birth, and Type 3 is the most severe form with multiple fractures and multiapical limb deformities. Cases which do not fit in any of the above are classified into type 4.[11-14]

The approach for the management of these cases is multidisciplinary, including bracing, physiotherapy, medical, and surgical intervention.[15,16] Intramedullary fixation of long bones aims to prevent the fractures and progression of deformities in these patients, with a view to maximize the motor development and improve the functional outcomes.[17-20] Rates of fractures were once as high as 1.5 fractures per patient per year with conventional treatment without stabilization.[21]

Delay in the presentation and surgical intervention is not unusual in OI, especially

How to cite this article: Bhaskar AR, Khurana D. Results of rodding and impact on ambulation and refracture in osteogenesis imperfecta: Study of 21 children. Indian J Orthop 2019;53:554-9.

Address for correspondence:
Dr. Atul R Bhaskar, Childrens Orthopaedics Surgical Services, Mhada Building 18, Off Link Road, Oshiwara, Mumbai, Maharashtra, India.
E-mail: arb_25@yahoo.com

Access this article online
Website: www.ijoonline.com
DOI: 10.4103/ortho.IJOrtho_202_18
Quick Response Code:

in those patients who hail from rural background due to various local and cultural beliefs. Most of the parents believe in a myth that plaster treatment is sufficient for fracture healing and deformity prevention. Repeated immobilization in refractures leads to muscle atrophy, decreasing bone strength, bone resorption, and refracture and a vicious cycle that eventually leads to poor functional outcome. Delayed surgery in this scenario is associated with prolonged rehabilitation, and therefore, repeat surgery for rod-related issues.

The aim of this study is to review the results of 21 OI patients with multiapical deformity in long bones who have been treated with intramedullary rodding and to record effects on ambulation and refracture rate.

Methods

A retrospective review of 44 OI patients treated between the years 2002–2016 was undertaken at our center. Only children with a minimum 2-year followup postintramedullary rodding were included. These children had complete records and regular followup. Children who underwent rodding but did not attend followup and physiotherapy were excluded from the study. Thus, only 21 cases were available for the final study. There were 11 boys and 10 girls, and with a mean age of 8.74 years (range 3–21 years) at the time of primary surgery. The clinical severity was based on the Sillence types-four patients were the Sillence type 1, seven were Type 3, and ten were of Type 4 [Table 1]. All children presented with fractures of long bones or preexisting multiapical angular deformities due to recurrent fractures. Five children had undergone previous surgeries, in three femoral plating was done, and two were treated with K wire, cerclage, and spica. In these 21 children, 35 femurs and 20 tibiae were operated in a staged manner to avoid significant blood loss. A single femur and tibia could be rodded in a single session. The standard Sofield and Millar's technique[20] of osteotomy, realignment, and intramedullary rod fixation was used in all the cases. Rush rods were used in stabilizing 28 femurs and eighteen tibiae, respectively. The indigenous Fassier-Duval Rod was used in eleven femur bones and 7 tibiae. In 2 femurs, with very narrow intramedullary canal, 2-mm K wires were used as rods. Supplementary plate fixation to control rotation was used in 5 children (6 femurs and 2 tibiae). All limbs were immobilized for an average duration of 6 weeks in a long leg cast. Four children also underwent humeral rodding and two being bilateral cases. Rush rods and F-D rods were used in four and two humeri, respectively. Eleven children had received intravenous bisphosphonates (BP) intermittently before the rodding procedure was undertaken.

Table 1: Sillence type of OI in study group

Age	Sex	Type of OI	Bones rodded		
			Femur	Tibia	Humerus
21	Male	4	2	2	
8.4	Female	4	2	2	1
9	Female	1	1		
9.2	Male	3	2	2	
5	Male	4	2	2	
8.2	Male	1	1		
6	Female	3	2	2	2
12.4	Male	1	1		
4	Male	4	2		
9	Male	4	2	2	
7.5	Male	3	2	2	
11	Female	3	1	1	
8	Male	4	2	1	
7.5	Female	4	2		
6	Male	4	2	1	1
9.6	Male	3	2		
9	Male	4	1	2	
12	Male	3	2		2
3	Female	4	2		
7.8	Female	3	1		
10	Male	1	1	1	

OI=Osteogenesis imperfecta

The preoperative ambulatory status was assessed by the H and B grading system,[22] [Table 2] and muscle power was recorded using the Medical Research Council (MRC) grade for major group of muscles, namely, hip abductors, hip extensors, quadriceps, hamstrings, and gastrosoleus complex. An average of five readings was taken as the final score. According to the H and B score, there were four household ambulators without aids (Grade 2A) with mean muscle power of MRC Grade 3. Four children were physiologic ambulators with aids (Grade 3B), with mean muscle power of MRC (Grade 3). Nearly 3 and 10 patients were wheel-chair independent (Grade 4) and three patients were carried by caregivers (Grade 5) with mean muscle power of MRC grade 2.7.

All children had serial radiographs taken to assess bone healing, and clinical assessment was undertaken at each review. Orthotics support was provided to stabilize the ligament laxity in children, and all parents and children were counseled for regular physiotherapy in the postoperative period.

At the final followup, ambulatory status of patients was recorded and graded according to the Hoffers and Bullock grading, and related complications were assessed.

Results

The mean followup duration was 34 months (range 24–48 months) after the index procedure. The mean time to osteotomy healing was 8 weeks (range 6–12 weeks) for both tibia and femur. Sixty six lower limb bones (41 femur and 25 tibia) and six humeri were operated by the standard Sofield-Millar procedure in a total of 21 OI patients. Staged surgery was done, at least 3 months apart, in cases with multiple bone involvement [Table 3].

Ambulatory status

At the final assessment, two of the four household ambulators (IIa) improved: one child improved to walking in community without aid (Ia) and one walking with aid (Ib), and 2 remained unchanged. Of the four physiological ambulators (Grade 3B), one child achieved community ambulation with orthosis (Grade 1A) and one improved to household ambulation with aids (Grade 2B). [Figure 1a-e]. Two children continued as physiological ambulators and remained unchanged. In the ten children who were wheelchair independent (Grade 4), seven improved in their ambulatory status. Five achieved physiologic ambulation with aids (Grade 3B) and two children improved to household ambulation with aids (Grade 2b). The remaining three children that were wheelchair dependent, two remained unchanged in their ambulatory status and one improved to physiological ambulation with aids (Grade 3b). Thus, after long bones stabilization, mobility improved in 11 of the 21 children (52% patients), and none of the preoperative ambulators deteriorated to nonambulatory grade. [Figure 2a-c].Before surgery, 13 children (61%) were confined to the wheel chair, and at the end of treatment only six (28%) remained dependent on the caregivers [Tables 4 and 5].

Table 2: Hoffer and Bullocks Grading for ambulatory potential

Grade	Description
Grade 1	Community ambulation (a) without aids, (b) with aids
Grade 2	Household ambulation (a) without aids, (b) with aids
Grade 3	Physiologic ambulation (a) without aids, (b) with aids
Grade 4	Wheelchair independent
Grade 5	Wheelchair sitter
Grade 6	Recumbent

Refracture

Sixty six femur and tibia were rodded in 21 children. Four children presented with distal femur fracture (six femurs), as the bone length increased with growth and the lower third femur was left unsupported without a rod. Both these children underwent exchange nailing. In three children (2 femurs and one tibia), the osteotomy site had incomplete union but none of the children experienced any pain with daily activities. In the upper limb, one child (one humerus) presented with nonunion for which repeat rodding and bone grafting was done as an additional procedure.

Before rodding, the refracture rate was 2.3 fractures per child every year. Following surgery, there was no child sustained fractures in the stabilized segment of the bone. At present, we advise rush rod revision when >20% of the bone segment remains unsupported either due to rod migration or natural growth. Revision rate is lower for telescoping nails, unless the male rod backs out or the device fails to elongate.

Infection

None of our children presented with any wound or deep infection in the followup period.

Table 3: Analysis of 21 osteogenesis imperfecta children in the study

Preoperative ambulation grade	Followup (months)	Ambulation grade at final followup	Sillence grade	MRC grade pre/postoperative	Duration of osteotomy healing (weeks)	Complications
4	48	2b	4	2/4	7	
2a	72	1b	1	3/4	8	
3b	50	1a	4	3/3	7	Refracture
4	48	3b	3	3/3	7	
3b	36	2b	4	2/4	7	
2a	36	2a	1	3/4	7	
4	38	4	3	3/3	6	
3b	26	3b	1	4/4	6	Refracture
4	26	3b	4	4/3	8	
2a	38	1a	4	4/4	8	
3b	48	3b	3	3/3	8	Refracture
5	56	5	3	3/3	7	
4	40	3b	4	3/4	6	
2a	56	2a	1	4/4	7	Rod migration
4	55	3b	4	3/4	8	
4	40	3b	3	3/3	7	
5	70	5	4	2/2	6	
4	60	4	3	2/4	7	
4	48	2b	4	3/4	7	
4	24	4	3	3/3	7	
5	18	3b	4	2/2	8	Refracture

MRC=Medical research council grading

Bisphosphonate treatment

Eleven children (52%) received intermittent bisphosphonate medication. Six children had received five cycles of intravenous pamidronate, and four were administered zolendronic acid annually over 4 years before the rodding. [Figure 3a-d].These injections were administered by respective physicians despite progressive bone deformation and recurrent fractures. In the followup period, the children continued to receive zoledronic acid once a year, after the osteotomies had healed.

Discussion

Children with untreated OI present with a classical appearance in their deformities on the lower limb. There

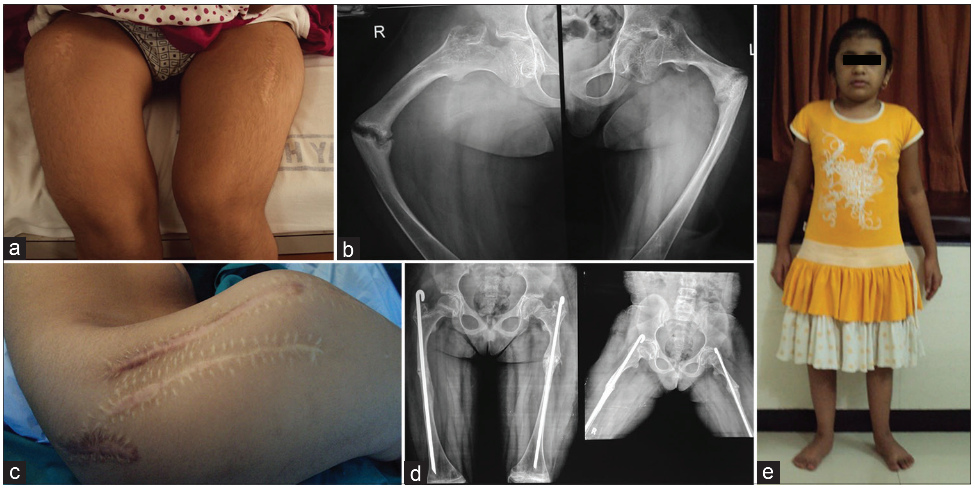

Figure 1: (a) A 12-year-old girl, H and B Grade IIIB, physiologic ambulatory, (b) bilateral femur fracture with deformity, (c) multiple scars of previous surgery: plating and "K" wire fixation done, (d) bilateral rush rod stabilization, (e) achieved community ambulation with orthosis (H and B Grade IB)

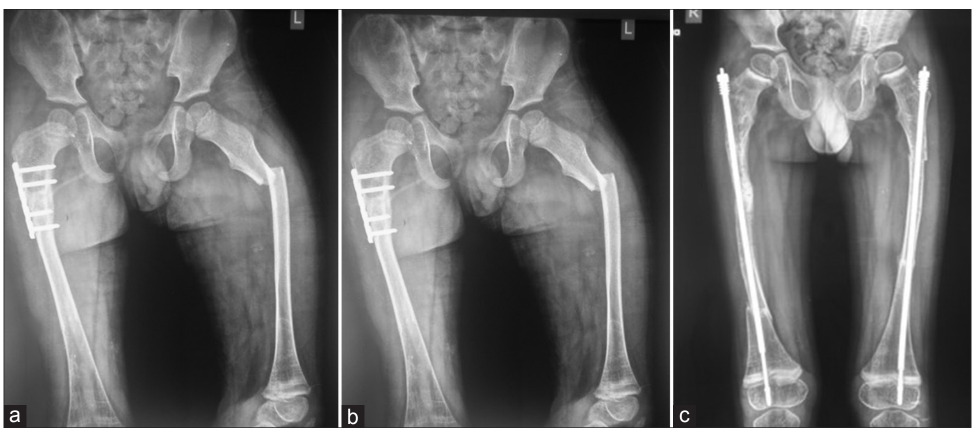

Figure 2: (a) A 7-year-old boy, H and B Grade IIIB, sustained pathological fracture of both femurs, (b) child underwent plating of both femurs, (c) child had fractured below the plate and underwent Fassier-Duval rodding. Achieved Grade IB ambulation

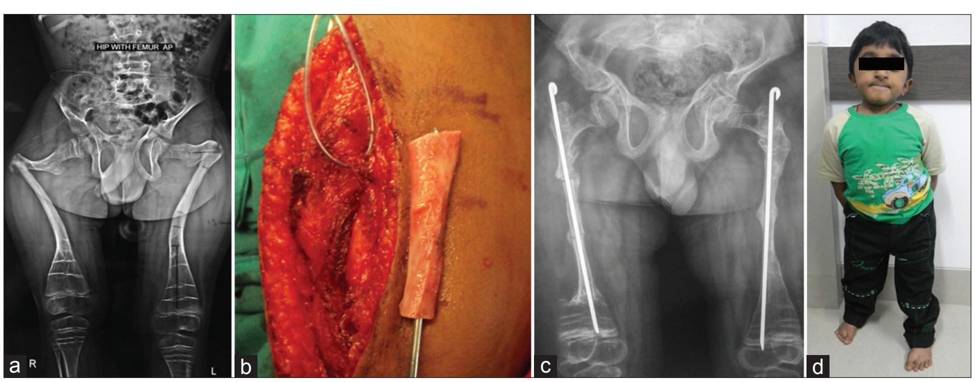

Figure 3: (a) A 9-year-old boy, H and B Grade IV, had received five cycles of bisphosphonates, (b) very narrow canal 2-mm k wires inserted, (c) Bilateral K wires inserted after multiple osteotomies, (d) achieved community ambulation with orthosis

Table 4: Change in Ambulatory status after rodding in 21 children

H and B grading	Preoperative number	At final follow up
1a	0	2
1b	0	1
2a	4	2
2b	0	3
3a	0	0
3b	4	8
4	10	3
5	3	2
6	0	0

H and B=Hoffer and Bullock

Table 5: Sillence type versus improvement in Hoffer and Bullock's grade in ambulation

H and B grade	Sillence type (*n*)							
	Sillence type 1		Sillence type 2		Sillence type 3		Sillence type 4	
	Pre	Post	Pre	Post	Pre	Post	Pre	Post
1a		1				1		
1b		1						
2a	3	2					1	
2b						1		2
3a								
3b	1				1	2	2	6
4					5	3	5	
5					1		2	2
6								

H and B=Hoffer and Bullock, *n*=number of children

is anterior and lateral bowing of the midfemur with proximal neck and subtrochanteric area in flexion, varus, and external rotation. The distal third of the femur is also in flexion and varus alignment. In tibia, the deformity is almost always an anterior bowing, and equinus at the ankle due to calf tightness. These characteristic deformities occur due to the muscle forces acting on the weak and fractured bone coupled with improper or inadequate treatment of the initial fracture. This leads to a vicious cycle of fracture and refracture causing progressive bowing deformities, muscle atrophy, and loss of ambulatory potential. Remodeling of these repeat fractures impacts the bone shape and density. There is sclerosis and cortical thickening on the concave size and thinning and stress shielding on the convex side making fixation with conventional plasters and wires untenable. Furthermore, in severe cases of limb deformities, it is difficult to judge the true limb alignment and intramedullary canal diameter of both femur and tibia.

Early rodding is thus favored to align the bone, improve lever arm for the muscles and prevent refractures. It helps to preserves muscle strength and prevents disuse atrophy due to repeated immobilization. We did, however, use postoperative casting for 4 weeks followed by lightweight orthosis to control joint laxity and allow early weight bearing. The youngest child was 3-year-old and underwent rodding of both femurs due to recurrent fractures. She was earlier confined to the wheel chair (Grade 4), and following surgery could walk with aids (Grade 2b). The family migrated to another country but remained in contact through E-mail and video call. Porat *et al*. stated that the Sillence type of patient does not necessarily reflect his or her potential for the postoperative ambulation.[17] Four children had mild OI (Sillence Type I), and none were community ambulators. Thirteen children in this study were Sillence Type 3 and 4 of which, eight (61%) improved in ambulatory status. The ambulatory potential was reflection of the preexisting deformity due to recurrent fractures, muscle weakness, and ligamentous laxity. Muscle power was measured for four main groups of muscles: hamstrings, quadriceps, hip abductors, and hip extensors, both in the preoperative and at final followup. There was an increase in muscle strength in the postoperative phase which gradually increased at final followup. We feel this improvement is due to functional activities, alignment of the bones, and the prevention of refractures after rodding. No comparison in muscle strength with Sillence type was done as there were few children in each group. The mean preoperative muscle strength was MRC grade 2.95 (range 2–4). We feel that this preoperative MRC grade was not a true reflection of muscle power as preexisting deformities, bone malalignment, and stress fracture-induced pain can preclude accurate assessment.

In this study, we used Hoffer and Bullock's[22] grading for the final evaluation of ambulation status. The postoperative muscle strength improved to MRC grade 3.42 for the entire group. The improvement in ambulatory potential was satisfactory in the present study group. Thirteen (62%) children were unable to walk before the rodding, but at 1 year followup, only six (28%) remained confined to the wheel chair. As per the H and B grading, ambulatory status changed in 12 (58%) after surgery, and proper rehabilitation increased. Our results are comparable to those stated by Porat *et al*.[17] in their study. Marafioti and Westin[23] reported excellent results in their series with 14 of 16 patients attaining ambulation after surgery. They used only two categories to classify ambulation potential, i.e., ambulatory and nonambulatory.

In our followup of 34 months, we reported only 2 cases of refracture in 21 cases, which is significantly less as compared to the refracture rate reported in the literature. Gerber *et al*.[21] have reported 1.5 fractures per year per OI patients which were treated with conservative means.

The probability that an OI child will walk depends on many factors, of which surgical intervention is one of them. Our main focus is early surgical intervention, as this decreases the duration of immobilization, leads to early initiation of rehabilitation program to gain muscle strength, thereby increases the chances of independent walking in near future.

There are few drawbacks of the study. We had a drop-out rate of 52% as many children from out of city had inconsistent followup. Many parents stop bringing their children after initial surgery due to the misconception that only one-time fracture treatment is necessary. Furthermore, we could not perform any statistical corelation due to very small number in each H and B group. A multicenter study can perhaps help us to understand the impact of rodding on function and ambulation.

OI remains a complex condition. While rodding can correct the bony deformities to restore bone and muscle alignment, it is the complex interplay of collagen in bone and soft tissues that determine the overall functional activity of the child. Future studies in improving the soft-tissue stability will become as important as surgery on the bone.

Conclusions

Intramedullary rodding treatment for recurrent fractures in children with OI improves their mobility potential. It also and prevents repeated cast application, disuse wasting, and osteopenia which can lead to deterioration in the quality of the long bones.

Declaration of patient consent

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

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Sykes B, Ogilvie D, Wordsworth P, Wallis G, Mathew C, Beighton P, *et al.* Consistent linkage of dominantly inherited osteogenesis imperfecta to the type I collagen loci: COL1A1 and COL1A2. Am J Hum Genet 1990;46:293-307.
2. Paterson CR. Osteogenesis imperfecta and other heritable disorders of bone. Baillieres Clin Endocrinol Metab 1997;11:195-213.
3. McLean KR. Osteogenesis imperfecta. Neonatal Netw 2004;23:7-14.
4. Rowe DW, Shapiro JR. Osteogenesis imperfecta. In: Avioli LV, Krane SM, editors. Metabolic Disease and Clinically Related Disorders. San Diego: Academic Press; 1998. p. 651-95.
5. Sousa T, Bompadre V, White KK. Musculoskeletal functional outcomes in children with osteogenesis imperfecta: Associations with disease severity and pamidronate therapy. J Pediatr Orthop 2014;34:118-22.
6. Sillence DO, Senn A, Danks DM. Genetic heterogeneity in osteogenesis imperfecta. J Med Genet 1979;16:101-16.
7. Van Dijk FS, Pals G, Van Rijn RR, Nikkels PG, Cobben JM. Classification of osteogenesis imperfecta revisited. Eur J Med Genet 2010;53:1-5.
8. Barnes AM, Carter EM, Cabral WA, Weis M, Chang W, Makareeva E, *et al.* Lack of cyclophilin B in osteogenesis imperfecta with normal collagen folding. N Engl J Med 2010;362:521-8.
9. van Dijk FS, Cobben JM, Kariminejad A, Maugeri A, Nikkels PG, van Rijn RR, *et al.* Osteogenesis imperfecta: A review with clinical examples. Mol Syndromol 2011;2:1-20.
10. Warman ML, Cormier-Daire V, Hall C, Krakow D, Lachman R, LeMerrer M, *et al.* Nosology and classification of genetic skeletal disorders: 2010 revision. Am J Med Genet A 2011;155A:943-68.
11. Gajko-Galicka A. Mutations in type I collagen genes resulting in osteogenesis imperfecta in humans. Acta Biochim Pol 2002;49:433-41.
12. Roughley PJ, Rauch F, Glorieux FH. Osteogenesis imperfecta – Clinical and molecular diversity. Eur Cell Mater 2003;5:41-7.
13. McEwing RL, Alton K, Johnson J, Scioscia AL, Pretorius DH. First-trimester diagnosis of osteogenesis imperfecta type II by three-dimensional sonography. J Ultrasound Med 2003;22:311-4.
14. Kocher MS, Shapiro F. Osteogenesis imperfecta. J Am Acad Orthop Surg 1998;6:225-36.
15. Shi CG, Zhang Y, Yuan W. Efficacy of bisphosphonates on bone mineral density and fracture rate in patients with osteogenesis imperfecta: A systematic review and meta-analysis. Am J Ther 2016;23:e894-904.
16. Abulsaad M, Abdelrahman A. Modified sofield-millar operation: Less invasive surgery of lower limbs in osteogenesis imperfecta. Int Orthop 2009;33:527-32.
17. Porat S, Heller E, Seidman DS, Meyer S. Functional results of operation in osteogenesis imperfecta: Elongating and nonelongating rods. J Pediatr Orthop 1991;11:200-3.
18. Brunelli PC, Frediani P. Surgical treatment of the deformities of the long bones in severe osteogenesis imperfecta. Ann N Y Acad Sci 1988;543:170-9.
19. Joseph B, Rebello G, Kant BC. The choice of intramedullary devices for the femur and the tibia in osteogenesis imperfecta. J Pediatr Orthop B 2005;14:311-9.
20. Sofield HA, Millar EA. Fragmentation, realignment and intramedullary rod fixation of deformities of the long bones in children: A ten-year appraisal. J Bone Joint Surg Am 1959;41:1371-91.
21. Gerber LH, Binder H, Weintrob J, Grange DK, Shapiro J, Fromherz W, *et al.* Rehabilitation of children and infants with osteogenesis imperfecta. A program for ambulation. Clin Orthop Relat Res 1990;257:254-62.
22. Hoffer MM, Bullock M. The functional and social significance of orthopedic rehabilitation of mentally retarded patients with cerebral palsy. Orthop Clin North Am 1981;12:185-91.
23. Marafioti RL, Westin GW. Elongating intramedullary rods in the treatment of osteogenesis imperfecta. J Bone Joint Surg Am 1977;59:467-72.