Case Series

Zahedan Journal of Research in Medical Sciences

Journal homepage: www.zjrms.ir

# Treatment Concepts of Osteogenesis Imperfecta

**Ramji Lal Sahu**

Department of Orthopedics, Sharda University, India

***Article information***

Article history:
Received: 30 Jan 2011
Accepted: 9 Apr 2011
Available online: 29 Dec 2011

Keywords:
Intramedullary nails
Correction
Osteogenesis imperfect

*Corresponding author at:
Department of Orthopedics,
Sharda University, India.
E-mail:
drrlsahu@gmail.com

***Abstract***

***Background*:** To explore the Application of the intramedullary nails for correction of deformity in the lower limbs and decrease the opportunity of refractures in children with osteogenesis imperfecta.

***Materials and Methods*:** From July 2005 to July 2009, 11 patients (5 males and 6 females), were recruited from Emergency and outpatient department having deformities of osteogenesis imperfecta in lower limbs. With 3 femurs and 5 tibias with deformity in lower limps were corrected by multiosteotomy and fixed with intramedullary interlocking nails, 6 (3 femurs and 3 tibias) for Rush nails; 6 (2 femurs and 4 tibias) for Ender nails; and 12 (6 femurs and 6 tibias) for flexible intramedullary nails. All patients were operated under general or spinal anesthesia.

***Results*:** All deformities were perfectly corrected. All patients were available at final follow up, for 9 months to 36 months, mean 18 months. 2 patients had delayed union, 2 had superficial infection in the incision or pin tract, and 1 had refractures postoperatively. The results were excellent in 72.727% and good in 27.272% patients.

***Conclusion:*** Multiosteotomy and fixed intramedullary nails can correct the deformity in the lower limbs perfectly and decrease the opportunity of refractures in children with osteogenesis imperfecta, which has been proved to be a reliable method.



## Introduction

Osteogenesis imperfecta, was described as a syndrome caused by a change on the connective tissue involving type-I collagen, which is the organic component of the bone [1]. However, with the recent DNA studies, it was proven that many patients with Osteogenesis Imperfecta did not show changes on the genes codifying collagen production [2]. Today, the concept has been broaden, being defined as a syndrome caused by a genetic change and with variable complexity levels. Diagnosis is pretty much provided by clinical examination and X-ray tests.

The classification by Sillence [3], published in 1979, has been employed for grouping these patients. The orthopedic treatment, both for fractures and for deformities correction, which consists of corrective osteotomy and stabilization of those bones, using several kinds of implant materials for osteosynthesis has been used by a number of authors [4-9]

Sofield and Millar [4] described a procedure for treating Osteogenesis Imperfecta, which consisted of multiple osteotomies and bone realignment using an intramedullary nail extending between both metaphyses of a same long bone. This procedure using fixed (non-extensible) nails allows long bones growth to "create" bone segments without nail protection, which occasionally presents fractures or deformities requiring new surgeries. Despite of this complication, this has been accepted as a preventive treatment for fractures and deformities. The objective of this study is to review patients diagnosed with Osteogenesis Imperfecta treated by using the Sofield technique, assessing surgical treatment of fractures and deformities on lower limbs in order to determine the effectiveness of the technique using fixed (non-extensible) nails.

## Materials and Methods

This prospective study was carried out at Orthopaedics department of M. M. Medical College from July 2005 to July 2009. It was approved by institutional medical ethics committee. A total of 11 patients with Osteogenesis Imperfecta submitted to multiple osteotomies and bone realignment with intramedullary nails on lower limbs reviewed at our institute were included in present study. A written informed consent was obtained from all the patients; they were explained about treatment plan, cost of operation, and hospital stay after surgery, and complications of anesthesia. They were followed up after surgery, and were clinically and radiologically assessed for fracture healing, joint movements and implant failure. According to the criteria the results are graded as excellent when the fractures unites within 16 weeks without any complications, good when union occurs within 24 weeks with treatable complications like superficial infection and knee stiffness and poor when union occurs before or after 24 weeks with one or more permanent complications like infection (osteomyelitis), implant failure, non-union, limb shortening and permanent knee stiffness. Delayed union was recorded when the fracture united between three to six months while nonunion was noted when union had not occurred after eight months of treatment. Follow-up was done.

The 11 patients (five boys and six girls) were classified according to the criteria described by Sillence [3]. Type IA (3), Type IB (1); Type II (0); Type III (5); Type IVA (0); Type IVB (2).(Table 1). Type I is the most prevalent form, including the milder bone weakness forms, with few fractures, no significant deformities and with normal height. We had four patients, three of whom in IA, with normal dentinogenesis, and one patient in IB com dentinogenesis imperfecta. Type II includes the most life-threatening kind, with a significant incidence of death at birth, with serious bone weakness, in which a number of intrauterine and delivery fractures can occur.

Death usually occurs during delivery or on the first days after birth. We had no patients included in this group. Type III includes classic cases showing significant bone weakness and deformity; the patients are usually short and show dentinogenesis imperfecta. We had five patients included in this group. Type IV includes patients with bone weakness, normal sclera, and skeletal deformity with short heights, which is subdivided into IVA with normal dentinogenesis and IVB, with dentinogenesis imperfecta. The two patients we had were included in the IVB type group. The mean follow up was 18 months (range 9 to 36 months). Inclusion criteria: 1.Children was within 3-15 years of age, 2.Only the lower limbs were included in this study Exclusion criteria: Upper limbs, spinal, and other extra osseous deformities were excluded in this study. On the 11 patients, a total of 32 nails were inserted in lower limbs. The procedures were carried out on 14 femurs (18% - six right and eight left femurs), and on 18 tibiae (81.9% -10 left and eight right tibiae). In this case series, three patients were submitted to tibial and femoral surgeries simultaneously (Fig. 1) Thirty-two fixed nails were employed, of which 9 were Interlocking nails, 9 for Rush nails, 6 for Enders nails and 10 for flexible intramedullary nails.

The preoperative and postoperative gait ability was assessed by dividing the patients into two groups of ambulating and no ambulating. (Fig 2 & 3) All the patients were no ambulating preoperatively, but after the operation they were being ambulated. The surgical procedure was carried out by using the same technique as described by Sofield and Millar4, which consists of exposing long bones and preserving periosteum, performing multiple bone osteotomies between proximal and distal metaphyses using an electric saw, achieving bone straightening. Prophylactic antibiotic therapy with cephalosporin (50 mg/Kg/day) was routinely used on anesthetic induction and on the first postoperative day. All patients had their operated limbs immobilized by plastered devices and/or removable immobilizers with a mean union time of 42 days (ranging from 40 to 51 days).

Ethical and legal procedure

The protocol was approved by an ethics committee and thus meets the standards of the Declaration of Helsinki in its revised version of 1975 and amendments made to it in 1983, 1989 and 1996 (JAMA 1997; 277:925–6).

**Figure 1.** Preoperative radiograph showing rotational and angular deformity of the tibia in severe osteogenesis imperfect

**Figure 2.** Postoperative, 3 years of follow – up, correction of deformity of both the femurs and tibiae

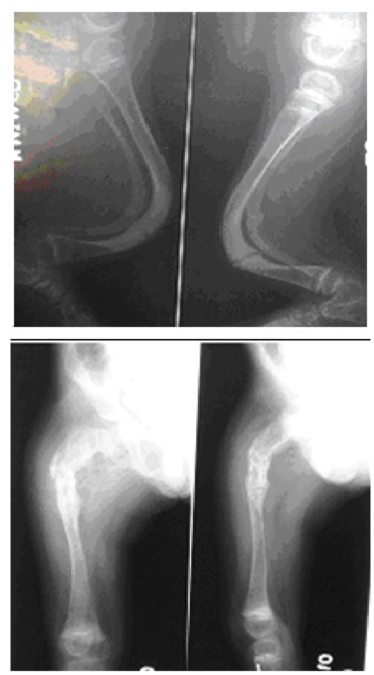

**Figure 3.** Preoperative radiograph of lower limb showing deformity in osteogenesis imperfecta.

**Table 1.** Osteogenesis imperfecta showing age, sex, classification by sillence and status of nails in the limbs

| Case | Age (Years) | Sex | Classification | Right tibia | Left tibia | Total tibia | Right femur | Left femur | Total femur | Total nails |
|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 3 | M | III | 1 | 1 | 2 | 1 | 1 | 2 | 4 |
| 2 | 5 | F | III | 1 | 1 | 2 | 1 | 0 | 1 | 3 |
| 3 | 7 | M | III | 1 | 1 | 2 | 1 | 0 | 1 | 3 |
| 4 | 9 | M | 1A | 0 | 1 | 1 | 0 | 1 | 1 | 2 |
| 5 | 11 | F | IVB | 1 | 1 | 2 | 0 | 1 | 1 | 3 |
| 6 | 7 | F | III | 1 | 1 | 2 | 1 | 1 | 2 | 4 |
| 7 | 5 | F | IB | 0 | 1 | 1 | 0 | 1 | 1 | 2 |
| 8 | 4 | M | 1A | 0 | 1 | 1 | 0 | 1 | 1 | 2 |
| 9 | 3 | F | 1A | 1 | 0 | 1 | 1 | 0 | 1 | 2 |
| 10 | 12 | F | III | 1 | 1 | 2 | 1 | 1 | 2 | 4 |
| 11 | 14 | M | IVB | 1 | 1 | 2 | 0 | 1 | 1 | 3 |
| Total | | | | 8 | 10 | 18 | 6 | 8 | 14 | 32 |

## Results

All the 11 children with osteogenesis imperfecta with deformity recovered fully, 3 of them in 6 months, 4 at 3 months and 4 at 4 months. All the children were unable to ambulate prior to surgery. They started standing and walking between 1-2 years postoperatively. There were residual deformity in 5 children but they managed to walk in few weeks.

The mean age at the primary procedure for nail insertion was eight years and five months, ranging from three years and to 14 years. The mean follow-up time for the first review was two years and one month (ranging from one year and five months to two years and seven months). In our series all the children took more time to unite the bone due to osteoporosis. In case number one, a male child of three years of age had deformity in both the tibia and femur were treated with multiple osteotomy to correct the deformity then fixed with elastic stable intramedullary nailing. There were coax vara in both the femur and angular deformity in both the tibia. After 2 months of postoperative day when child started walking, there was refracture in right lower tibia at osteotomy site; it took 4 months time to unite. In case number 6 and 10, were similar to case number one was treated similarly with osteotomy and flexible nails. There were no refracture in these cases.

Case number 2 and 3 were treated with Enders nails. Case number 7 and 8 showed infection superficially but were cured with antibiotics within a week. Case number 4, 5, and 11 were treated with interlocking nails after corrective osteotomy. Case number 7, 8 and 9 were treated with rush nails. Delayed unions in left tibia were seen in case number 7and 8. Concerning osteotomy complications with the use of non-extensible nails: none of the patients was submitted to blood transfusion. Osteotomies showed a union rate of 100%. In the present study, the procedure has been shown not to interfere with physis, because we had no patient with physeal changes. We didn't find growth disorders inherent to the adopted procedure. (Table 2 & 3) .In a subjective measure of outcome at follow-up, 8 (72.727%) of the patients were excellent and 3 (27.272%) good; no patients or parents reported their out-come as not satisfied. At follow-up all patients went on to osseous union and regained a full range of movement after rehabilitation.

**Table 2.** Results of Osteogenesis Imperfecta, showing union time and complications of different Implants (n=11)

| Case | Age | Type of implant used | Time to unite | Complications |
|---|---|---|---|---|
| 1 | 3 | Elastic nail | 6 months | Refracture |
| 2 | 5 | Enders nail | 4 months | Nil |
| 3 | 7 | Enders nail | 4 months | Nil |
| 4 | 9 | Interlocking nail | 3 months | Nil |
| 5 | 11 | Interlocking nail | 4 months | Nil |
| 6 | 7 | Elastic nail | 3 months | Nil |
| 7 | 5 | Rush nail | 6 months | Delayed union,Superficial infection |
| 8 | 4 | Rush nail | 6 months | Delayed union,Superficial infection |
| 9 | 3 | Rush nail | 4 months | Nil |
| 10 | 12 | Elastic nail | 3 months | Nil |
| 11 | 14 | Interlocking nail | 3 months | Nil |

**Table 3.** Results of Osteogenesis Imperfecta- Showing pre and post operativeambulation (n=11)

| Type | Patients number | Pre-operative | Post operative |
|---|---|---|---|
| IA | 27.2% | Non ambulating | Ambulating |
| IB | 9.2% | Non ambulating | Ambulating |
| III | 45.5% | Non ambulating | Ambulating |
| IVB | 18.1% | Non ambulating | ambulating |

**Figure 4.** Post operative radiograph of lower limb showing correction of deformity and fixation with flexible nail

## Discussion

The concept of multiple osteotomies and internal fixation with an intramedullary rod for deformity correction and prevention of long-bone fractures in patients with osteogenesis imperfecta was first described by Sofield and Millar [4] in 19591 and has been widely accepted. In 1963, Bailey and Dubow [10] introduced an elongating rod system, whereby a hollow sleeve and an internal obturator were engaged and anchored by T-pieces at the proximal and distal epiphyses of the long bones. We evaluated our patients who were classified according to the Classification by Sillence [3].

Our series encompassed types IA, IB III and IV B of the classification by Sillence. Type III had the highest number of patients (45.5%), Type IA (27.2%), Type IV (18.1%), and IB (9.2%) And Type II, for being the most life-threatening one showing the highest incidence of death at birth, included no patient. These data are consistent to the study by Ryoppy et al [11]. The procedure of conducting multiple osteotomies fixated with intramedullary nails in order to fix deformities and prevent recurrent fractures on patients with Osteogenesis Imperfecta has been accepted since 1959, when Sofield and Millar described this procedure. It has been modified by a number of surgeons [6, 9, 12-14], but its principle, which consists of multiple osteotomies, realignment and fixation with intramedullary nail on long bones, remains the same. This procedure improves the quality of life for these patients, although complications are commonly seen, such as bone segments deprived of nail protection due to bone growth, enabling the occurrence of fractures or recurrent deformities, nail migration, pseudoarthrosis and union delay [4, 13].

Indeed, with a continuous growth, bone segments exceeding the nail can be deprived of protection, favoring deformities and nail protrusion through the cortical. The bone can either fracture or deform at this level, requiring nail replacement by a longer one and a new realignment of the limb. In our study, none nails required review. The procedure is associated to a low morbidity rate. Osteotomies showed union in all patients, no blood transfusion was required, and no deep infection or anesthetic complications were found. Regarding the best time for surgery, literature does not indicate a consistent concept about the best age to fix deformities or the best time to prevent recurrent fractures. Ryoppy et al. recommend interventions in children using non-extensible nails, emphasizing that the early realignment and stabilization of the lower limbs improves motor development, with no minimum age for surgery [11]. Williams et al. concluded that the best time to start inserting nails is when deformities are detected [14]. Tiley and Albright reported that the optimal age for starting lower limbs correction is when the patient keeps orthostatism [8]. This study did not determine the best time for surgical procedure. The question is: what is best for the patient: to provide an early treatment before a deformity or fracture occurs, or to wait and treat the complications? We believe that a surgical treatment using non-extensible nails is indicated when complications such as fractures and/ or deformities are present, based on the fact that these nails do not follow bone growth, if early implanted, complications will eventually occur, resulting in a larger number of review surgeries. Elastic stable intramedullary nailing has several advantages [15]: The operation can be done through small incisions, without significant trauma to tissue; the reduction is performed as either a closed or an open procedure, thus the pathological bone area may be exposed and, if required, biopsied.

The fracture haematoma, the periosteum and the surrounding soft tissue are spared to encourage accelerated formation of callus; - The system is elastic and not stiff, thereby rapidly inducing callus with subsequent secondary fracture healing; - The metal nails can be left in place without negatively affecting the healing of the bone disease, since, according to the prevailing opinion in the literature, no negative influence on the growth of the affected bone has been demonstrated to date; - The removal of the implants is simple, quick and atraumatic.

Elastic nails have a useful role in the management of benign pathological fractures of long bones. The pathological fracture may be as a result of local bone weakness or more generalized pathology. Asymptomatic defects such as fibrous cortical defects or unicameral bone cysts can present as a fracture [16, 17]. In general, these fractures will unite and often cyst itself will heal. In our study assessing pre-and postoperative gait, we divided the patients as ambulating and non-ambulating. All the children were unable to ambulate prior to surgery. They started standing and walking between 1-2 years postoperatively. In summary, the surgical treatment of lower limbs deformities and fractures on Osteogenesis Imperfecta shows the following advantages: gait status is kept, does not present physical injury sequel, brief hospitalization time (mean of four days), and deformity fixation with improved limb function. One disadvantage would be bone growth enabling the emergence of areas without protection of the internal fixation, which might cause recurrence of deformities, fractures and wire migration. These complications usually require review surgeries.

## Conflict of Interest

No conflict.

***References***


1. Corrêa EA, Cunha TNA, Barbenio CAV and Oliveira MNR. Osteogênese imperfeita na síndrome de Down: Relato de um caso. Rev Bras Ortop 1992; 27: 180-2.
2. Assis MC, Plotkin H, Glorieux FH and Santili C. "Osteogênesis imperfecta": novos conceitos. Rev Bras Ortop 2002; 27: 323-7.
3. Sillence D. Osteogenesis imperfecta an expanding panorama of variants. Clin Orthop Relat Res 1979; (159): 11-25.
4. Sofield HA, Millar EA. Fragmentation, realignment and intramedullar rod fixation of deformities of the long bones in children. A Ten-Year Appraisal. J Bone Joint Surg Am 1959; 41: 1371- 91.
5. Bailey RW, Dubow HI. Studies of longitudinal bone growth resulting in an extensible nail. Surg Forum 1963; 14:455-8.
6. Williams PF. Fragmentation and rodding in osteogenesisimperfecta. J Bone Joint Surg Br. 1965; 47: 23-31
7. King JD, Bobechko WP. Osteogenesis Imperfect. An orthopaedic description and surgical review. J Bone Joint Surg Br 1971; 53: 72-89.
8. Tiley F, Albright JA. Osteogenesis imperfecta: Treatment by multiple osteotomy and intramedullary rod insertion. Report on 13 patients. J Bone Joint Surg Am 1973; 55(4): 701-3.
9. Marafioti RL, Westin GW. Elongation intramedullary rods in the treatment of osteogenesis imperfecta. J Bone Joint Surg Am. 1977; 59(4): 467-72
10. Bailey RW, Dubow HI. Studies of longitudinal bone growth resulting in an extensible nail. Surg Forum. 1963; 14: 455-8.
11. Ryoppy S, Alberty A, Kaitila I. Early semiclosed intramedullary stabilization in osteogenesis imperfecta. J Pediatr Ortop1987; 7(2): 139-44.
12. Middleton RW. Closed intramedulary rodding for osteogenesis imperfecta. J Bone Joint Surg Br 1984; 66(5): 652-5.
13. Porat S, Hiller E, Serdnan DS and Meyer S. Function results of operation in osteogenesis imperfecta. J Pediatr Orthop 1991; 11(2): 200-3.
14. Williams PF, Cole WHJ, Bailey RW, et al. Current aspect of the surgical treatment osteogenesis imperfecta. Clin Orthop Relat Res. 1973; (96): 288-98.
15. Knorr P, Schmittenbecher PP, Dietz HG. Elastic stable intramedullary nailing for the treatment of complicated juvenile bone cysts of the humerus. Eur J Pediatr Surg 2003; 13(1): 44-49.
16. Kaelin AJ, MacEwen GD. Unicameral bone cysts: natural history and the risk of fracture. Int Orthop 1989; 13(4): 275-282.
17. Ahn JL, Park JS. Pathological fractures secondary to unicameral bone cysts. Int Orthop 1994; 18(1): 20-22.


***Please cite this article as*:** Lal Sahu R. Treatment concepts of osteogenesis imperfecta. Zahedan J Res Med Sci (ZJRMS) 2012; 14(6): 42-46.