

Management of Lower Limb Deformities in Children with Osteogenesis Imperfecta

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Osteogenesis imperfecta (OI) often leads to severe lower limb (LL) deformities due to recurrent fractures that significantly hamper ambulation. We describe our management experience of correction of LL deformities in four children with OI. Medical management consisted of peri and postoperative pamidronate therapy, calcium supplementation and rehabilitative care. Deformities were corrected with multiple osteotomies and intramedullary fixation by titanium elastic nails. At a mean follow-up of 30 months, all children have significantly reduced fracture incidence and have no evidence of recurrence of deformities with improved ambulatory status. We emphasize the importance of combined medical and surgical therapy for these patients.

Key words: Bisphosphonates, Deformities, Management, Osteogenesis imperfecta.

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Osteogenesis imperfecta (OI) is a group of heterogenous disorders characterized by increased bone fragility, multiple fractures, deformities and osteopenia [1]. The treatment of lower limb deformities in OI prevails to be a therapeutic challenge requiring multidisciplinary management to improve function and quality of life [1,2]. Surgical correction of deformities and stabilization of long bones using an intramedullary fixation (IF) device has proven to be an effective measure [1,3]. However, data from India addressing these deformities in OI patients is scant. In this study, we describe our management experience using combined medical and surgical modalities with a good outcome.

METHODS

The case records of OI children presenting with lower limb deformities over a period of two years from June 2007 to June 2009 were retrieved.

Diagnosis of OI was based on clinical and radiological characteristics. Initial assessment included serum calcium, phosphorus, alkaline phosphate and vitamin D levels. Anteroposterior and lateral radiographs of lower limbs were performed for assessment of deformities. Additionally, bone mineral density was evaluated by dual energy X-ray absorptiometry (DEXA) of the lumbar spine. Pre and post therapy ambulatory status was graded according to the modified Hoffer and Bullock's classification [4]. The annual fracture rate was calculated based on historical and follow-up data. The conduct of the study was approved by the Hospital Ethics Committee.

All patients received medical (pre and postoperative) and surgical therapy. Preoperative management constituted calcium supplementation, supportive care and nutritional rehabilitation. Pamidronate was administered preoperatively at a dose of 1 mg/kg body weight for 3 days. Open

WHAT THIS STUDY ADDS?

- Combined medical and surgical modalities are feasible options in India for correction of lower limb deformities in children with osteogenesis imperfecta.

reduction and deformity correction by multiple osteotomies was performed followed by intramedullary fixation of implants by 'Seekh-Kabab' method. Intramedullary implants used were titanium elastic nails (TEN). The nails were inserted under C-ARM guidance and appropriate position was confirmed postoperatively using the same. Postoperatively plaster immobilization was done for 6 weeks. After the osteotomies united, ambulation with the aid of crutches was encouraged and patients were started on a rehabilitation programme. Pamidronate was continued at a dose of 1 mg/kg/day for 3 days in 3 monthly cycles starting 3 weeks after surgery for 1 year. Regular bone studies were performed after each pamidronate infusion. All cases were followed up clinically and radiologically regarding deformity correction, mobility status, re-fracture, infection, nail migration, breakage and bending of nails.

RESULTS

There were four male OI children with a mean age of 7.3 (range 6-10) years. All presented with unilateral or bilateral femoral and/or tibial bowing with history of multiple fractures after trivial trauma since early childhood and blue sclera. There were no other extraskkeletal manifestations. Family history was present in two of them. The ambulatory grade ranged from 3b to 4 (**Table I**). DEXA Z-score was suggestive of severe osteoporosis (range 5.68-3.01). In our series of four patients, the following deformities were surgically corrected: 1 bilateral femur with unilateral tibia, 1 bilateral femurs, 2 unilateral femur and unilateral tibia.

Radiological correction of angular deformities was good. In one, there was a cut-through of the tibia fixation requiring re-do surgery and fixation. The other complications encountered were backing out of femoral nails in 2 patients after 6 months to 1 year of surgery. The nails were re-positioned without any further problem. Bony reunion occurred in all. At a

mean follow-up of 30 months, the patients have reduced incidence of fractures (annual fracture rate of zero), better bone densities, no recurrence of deformities and improved quality of life (**Table I**). The children as well as parents are satisfied with their overall progress and improvement in ambulatory status.

DISCUSSION

Recent advents in medical and surgical therapy have led to low fracture incidence and improved function and quality of life in OI children [1,5]. However, there is limited information available regarding the medical and surgical management of lower limb deformities in children with OI from India.

As bisphosphonates do not cause resolution of pre-existing bony deformities, the preferred treatment of correction of lower limb deformities is IF [1,2]. Data pertaining to the surgical management of OI children is available from a single centre of India where bisphosphonates were not used [6]. There are reports addressing the effect of bisphosphonates on reduction in fracture rate in OI children from India [7,8]. However, none of them studied their effect in correction of LL deformities as an adjuvant to surgery.

The use of bisphosphonates can be substantiated by the fact that they reduce bone turnover and thereby, increase cortical bone density. As observed by us, bisphosphonates allow safer and more

TABLE I RESPONSE TO THERAPY IN 4 PATIENTS

Age (Yrs)	Follow up (Months)	Ambulatory grade	
		Pre therapy	Post therapy
6	40	4	3b
7	33	4	2b
6	37	3b	1b
10	20	3b	1b

AFR: Annual fracture rate.

efficacious surgical management of OI children and also reduces future incidence of fractures. The timing of bisphosphonate treatment relative to surgery is important and it may improve prognosis in severe forms [9]. In a study from Egypt, authors have advocated the role of combined approach (surgical and medical treatment) and both pre-operative and post-operative pamidronate for these patients [10].

We used TEN due to its easy availability and utility in IF. Other implants were not available and if available, the sizes were not suitable for younger children. Elongating nails allow for growth of the bone and decrease the number of repeat operations [11]. TEN is not an expandable nail. However, TENs were used to keep as an internal splint for a couple of years till the effects of medical treatment kicked in and prevented further fractures. Additionally, focus was laid on gentle physiotherapy initially, followed by muscle strengthening exercises and gait training later. The advantage of TEN lies in its flexibility which plays an important role in the correction of complex deformities. The disadvantage is absence of secure fixation at times leading to backing of the nail, as was observed in two cases.

Although having a short follow-up with a modest sample size, our experience with this combined medical and surgical management seems promising in terms of successful rehabilitation of these children in the future.

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