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# Surgical Treatment of Bilateral Tibia Deformity in a 9-Year-Old Child Suffering from Osteogenesis Imperfecta Type III: A Case Report

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

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**Conflict of interest:** None declared

**Patient:** Male, 9
**Final Diagnosis:** Osteogenesis imperfecta type III
**Symptoms:** Walking difficulties
**Medication:** —
**Clinical Procedure:** Osteotomies and realignment of bone deformity
**Specialty:** Orthopedics and Traumatology

**Objective:** Congenital defects/diseases

**Background:** Osteogenesis imperfecta is a rare inherited connective tissue disorder that is mainly characterized by long bone deformities and increased susceptibility to bone fractures. The aim of this study was to present a surgical technique in a child suffering from osteogenesis imperfecta and progressive, severe bowing deformity of both tibias, as an alternative method to advanced, innovative surgical systems.

**Case Report:** A 9-year-old child (male) was referred to our orthopedic clinic for inability to walk independently due to extreme anterior bowing of both tibias. After the diagnosis of osteogenesis imperfecta type III was established, bilateral surgical treatment with multiple osteotomies and intramedullary, flexible Titanium Elastic Nail System (TENS) nails was decided. Six months post-operatively callus formation was obvious in x-rays and at the latest follow-up (1 year post-operatively) the patient regained the ability to walk independently.

**Conclusions:** In types of osteogenesis imperfecta which are characterized by extreme deformities (type III) surgical intervention seems to be the only solution for these patients to walk again. We present a relatively simple technique of correcting such deformities, indicating that no matter which technique will be used (simple or more complicated) the final goal should be to restore the walking ability with as little complications as possible.

**MeSH Keywords:** Bone Malalignment • Disabled Children • Osteogenesis Imperfecta • Osteotomy

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## Background

Osteogenesis imperfecta (OI) is a term describing a group of connective tissue disorders, that are characterized by low bone mass and increased bone fragility (brittle bone disease) [1,2]. Orthopedic manifestations include frequent fractures, scoliosis, and long bone deformities that can severely influence the walking ability [3,4]. Extraskeletal manifestations of osteogenesis imperfecta can include blue/gray sclera, hearing loss, aortic root dilatation, dentinogenesis imperfecta, macrocephaly, and basilar invagination [5,6].

The mainstream of treatment in children who present with extreme deformities of their long bones consists of multiple osteotomies and load sharing intramedullary devices in order to re-align the bone and provide adequate stabilization [3–6]. The aim of our study was to present our surgical strategy, in a 9-year-old male patient with severe, progressive, bilateral tibia deformity suffering from OI, with osteotomies and intramedullary, flexible Titanium Elastic Nail System (TENS) nails, that can be an excellent alternative when doctors or hospitals do not have access to advanced surgical systems, such as telescoping rods.

## Case Report

A 9-year-old refugee male patient presented by an International Charity Organization to our department with progressive bilateral deformity of femur and tibia. The bowing of both tibias was so severe that walking was impossible (Figure 1A–1D). Regarding our patient's medical history, he had not received any appropriate treatment at all (no bisphosphonates treatment, no previous procedures). Over the years, he experienced extreme deformity of both tibias, leading to anteromedial deformity.

Clinical examination revealed bilateral genu varum. Range of motion of the ankle, knee, and hip joints was not limited. No tenderness was found in the knee and ankle joints and no limb length discrepancy was present. Pain was localized exactly over the apex of tibia deformity in both legs. Neurovascular examination was normal. The main problem of our patient was the loss of capacity for independent walking and upright standing. There was an absolute need for crutches.

Standing anteroposterior (AP) full limb radiographs of both lower extremities were taken. Radiographs of both tibias were also taken in the AP and lateral view. A significant bowing of both tibias and fibulas was revealed with the apex of curve directed anteromedially (Figure 2A–2D) as well as bowing and varus deformity of both femurs (Figure 2E, 2F). Low bone density, thickening of the cortical bone, medullary canal narrowing and synostosis between tibia and fibula were noticed (Figure 2A–2D). A complete blood test and laboratory analysis were performed. Except for a mild elevation of ALP (alkaline phosphatase), the laboratory evaluation was normal.

### Pre-operative planning

After discussion with the parents, operative treatment with simultaneous bilateral correction of both tibias was decided. The pre-operative plan included the diameter of the tibia bone, the narrowing of the medullary canal, the type of bowing-deformity, and the levels of the possible osteotomies. Radiographic evaluation of mechanical axis of the tibia, anatomical axis of the tibia, medial proximal tibial angle (MPTA), lateral distal tibial angle (LDTA), and joint line congruency angle (JLCA) in AP views were also measured. All these angles were found to be within the normal range. The pre-operative plan was based on the principles of deformity correction that have been well described in the literature [7–9]. The point of intersection of the proximal and distal axis lines of the deformed tibias was evaluated in order to find the center of rotation of angulation (CORA). Because in our patient the CORA was outside the boundaries of the involved bones, it was clear that a multiapical deformity was present, and more than one osteotomy would be required to achieve an acceptable alignment of the tibias (Figure 3). The final goal was to realign the axis of the legs, provide adequate stability in order to avoid an impending fracture, and to provide the patient with the ability to walk. Taking into account the age of our patient (open physes) and the fact that we had to choose an alternative to advanced surgical systems (because our patient had no health insurance as a refugee child presented to our clinic from a charity organization) we decided to proceed with multiple osteotomies and intramedullary devices: TENS nails, Synthes 3.5 mm diameter.

### Surgical technique

General anesthesia was administrated. The patient was placed in the supine position and under fluoroscopy, 2 TENS nails were inserted antegrade with the entry point being just distal to the proximal epiphyseal plate of the tibia, at the medial and the lateral side of the tibia tuberosity, respectively (Figure 3A, 3B). Insertion of these nails was performed until the level of the pre-planned (more proximal) osteotomy. An open approach directly over the curve of the deformity followed the insertion of the nails. Kirschner wires (K-wires) were used to mark the 2 levels of the osteotomies and to use them as joysticks to help reduction (Figure 4A, 4B). The osteotomies were prepared by performing multiple holes with the drill and finished using an osteotome. An osteotome (and not a saw) was used to gently break the cortical bone at the correct level with the aid of the pre-drilled holes. After performing the osteotomy, the bone was realigned (Figure 5A, 5B), however, more multiple small-sized additional osteotomies were



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**Figure 1.** (**A, C, D**) Clinical presentation of our patient. Bilateral severe bowing and deformity of the tibia due to osteogenesis imperfecta (OI). (**B**) The bowing of the tibia was so extreme that even walking was impossible.

needed (Figure 5C). The synostosis between the tibia and fibula was also removed (Figure 5C). Intra-operatively, it was crystal clear that the medullary canal was extremely narrowed at the level of the deformity. We proceeded with multiple drilling of the canal to increase the diameter (Figure 6A). Then, the 2 nails that were already inserted till the level of the proximal osteotomy, were passed through the 2 fragment ends, which were also stabilized with an additional K-wire (Figure 6B–6D). A significant step was the final adaptations and adjustments of the periosteum (Figure 7A, 7B). Figure 8 shows the post-operative x-rays of our patient.

## Follow-up

Both legs were immobilized in a below-knee cast for 6 weeks. Figure 9 shows our patient's tibia alignment at 15 days of follow-up. Weight-bearing was not allowed for 3 months. The patient used crutches for mobilization. Touch weight bearing was



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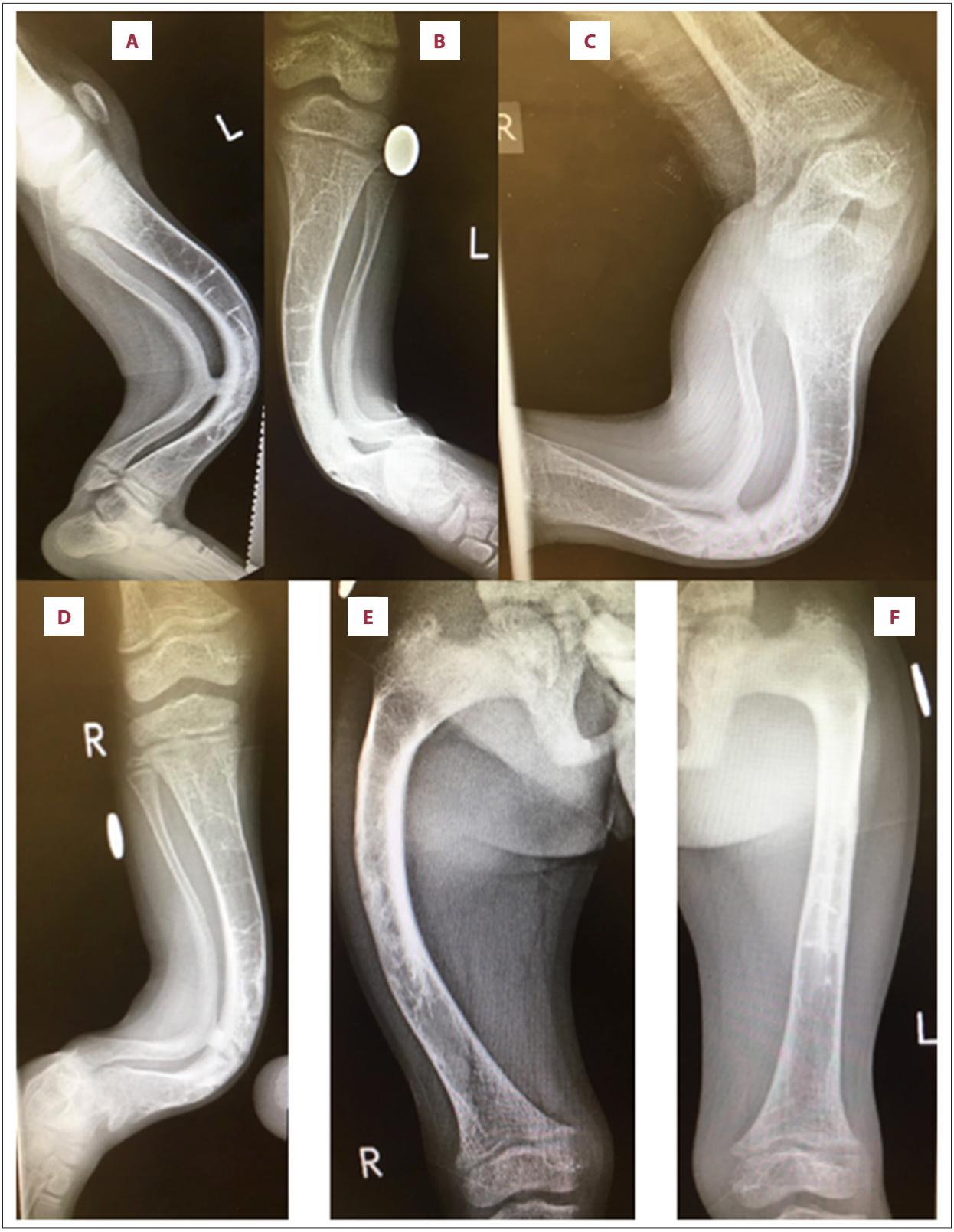


**Figure 2.** (**A–D**) F+profile views of the left and right tibia of our patient. Bowing of tibia, cortical thickening, narrow of the medullary canal, low bone density, and synostosis between tibia-fibula can be seen. (**E, F**) Anterolateral bowing and varus deformity of both femurs.



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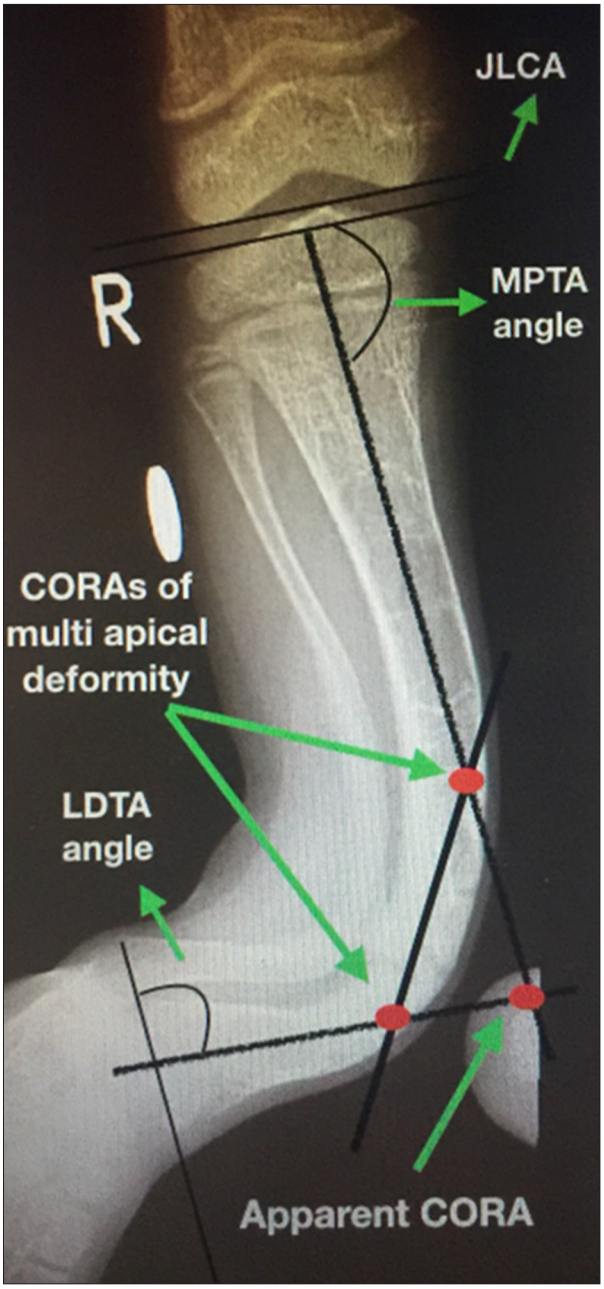


**Figure 3.** In our patient's AP right tibia x-ray, the center of rotation of angulation (apparent CORA) lies outside the boundaries of the bone and the point of obvious deformity (intersection of proximal and distal black lines). A multi apical deformity exists. Multiple osteotomies will be needed. MPTA, LDTA, and JLCA were also measured. The same planning was performed on the left tibia. AP – anteroposterior; CORA – center of rotation of angulation; MPTA – medial proximal tibial angle; LDTA – lateral distal tibial angle; JLCA – joint line congruency angle.

allowed at 3 months and full-weight bearing was not allowed until signs of callus formation were seen on x-ray, 6 months post-operatively (Figure 10). Complete radiographic union was established 9 months post-operatively. At 1-year follow-up, a small degree (10° to 15°) of tibia valgus was present but the patient was walking without pain, and range of motion of both knee and ankle joints was normal. No evidence of radiographic nonunion was observed. Regarding the femur deformity (bowing and genu varum) no surgical intervention was decided at the time, but close follow-up every 6 months was scheduled in order to proceed with surgical intervention at an appropriate time.

## Discussion

Osteogenesis imperfecta (OI) is a connective tissue disease characterized by a wide variety of phenotypic and molecular heterogeneity [10]. It is an unusual heritable disease (1 in 10 000 to 20 000) with 90% of patients having mutations in type I collagen genes (COL1A1 and COL1A2) [4,10]. The genetic defect is inherited either with autosomal dominant transmission or with autosomal recessive transmission [11].

The first classification system of OI into 4 types (I–IV) was made in 1979 by Silence et al. and it was mainly used for the clinical and radiological classification of OI: type I mild non-deforming, type II perinatal lethality, type III severely deforming, and type IV moderate deforming [10,12]. Since then, new genes have been discovered and the classification has been expanded with OI types V–VII mainly based on cases with unknown genetic etiology [5,13].

OI type I is mostly characterized by a 50% reduction of the amount of collagen type I (quantitative disorder in collagen) while OI types II–IV by sufficient but abnormal collagen I production (qualitative disorder in collagen) [5,10]. Low bone mass is one of the main characteristics of OI that leads to structural deficiency, but the mechanical quality of the bone material is also reduced [14]. This is no surprise as one of the main organic components of bone is collagen type I that is affected by the genetic defects. In addition, deformities of long bones (tibia, femur) and progressive scoliosis are common manifestations of OI [15]. Bowing of large bones result in extreme mechanical stresses in the apex of curvature and thus, they are a significant risk factor of bone fracture [6]. Our patient not only was "one step" before sustaining a fracture but he has already lost the walking ability due to progressive tibia deformity.

It can be easily understood, the importance of surgical realignment and stabilization of long bone deformities. In the literature, it seems that the use of load-sharing devices such as intramedullary Rush rods. Kuntscher rods, K-wires,



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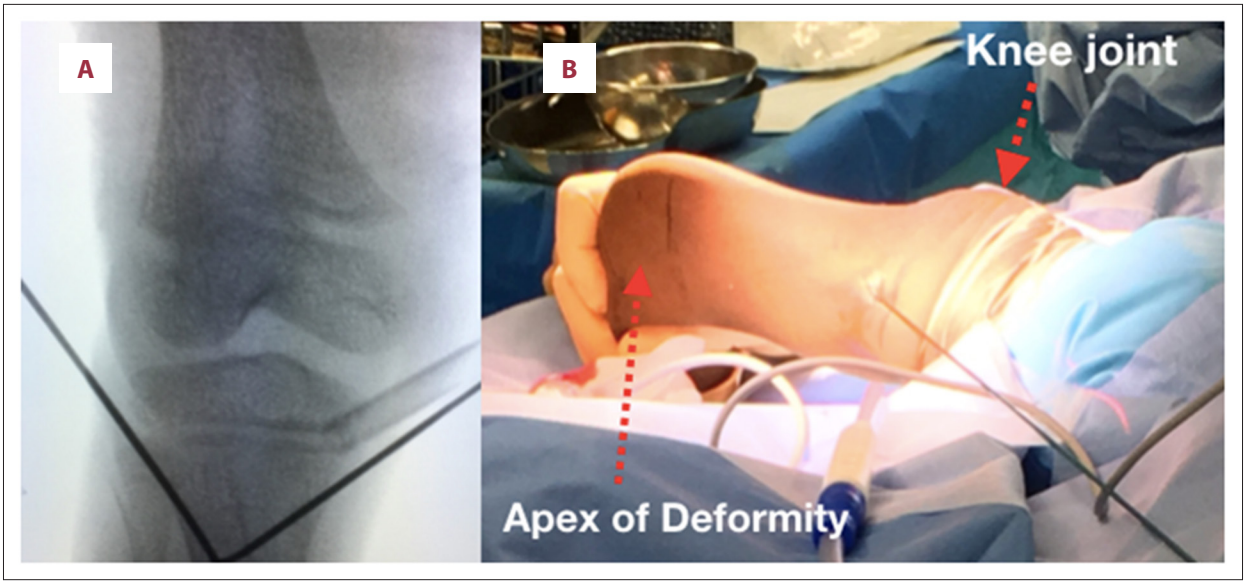


**Figure 4.** (**A**) Intra-operative x-ray showing the insertion of TENS nails distal to the epiphyseal plate, medial and the lateral to the tuberosity of the tibia. (**B**) Intra-operative picture while inserting the TENS nails. The red arrows show the tibia-bowing deformity and the knee joint. TENS – Titanium Elastic Nail System.

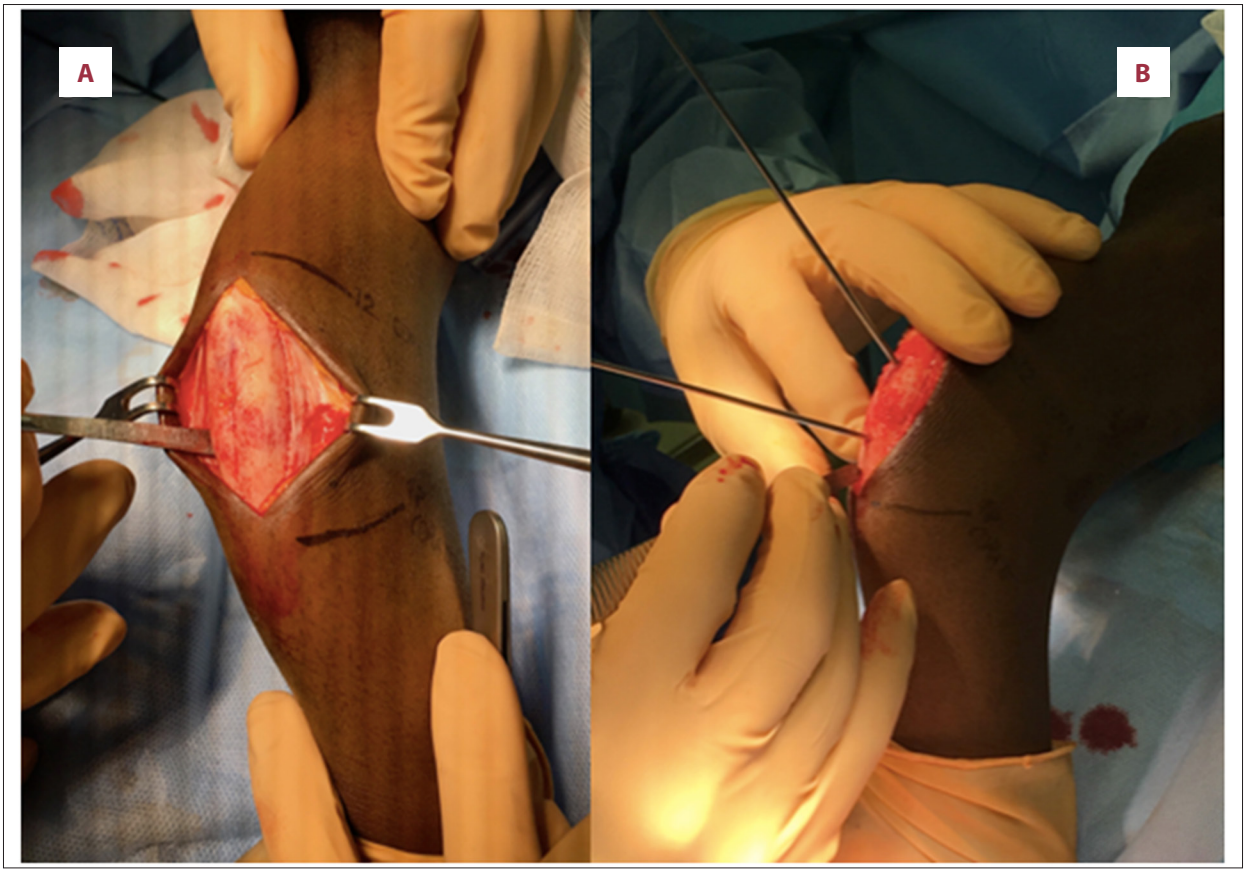


**Figure 5.** (**A**) Incision directly over the apex of the deformity. (**B**) K-wires were used to mark the levels of the pre-operative planned osteotomies. Both steps were performed after insertion of the nails until the level of the most proximal osteotomy. K-wire – Kirschner wires.



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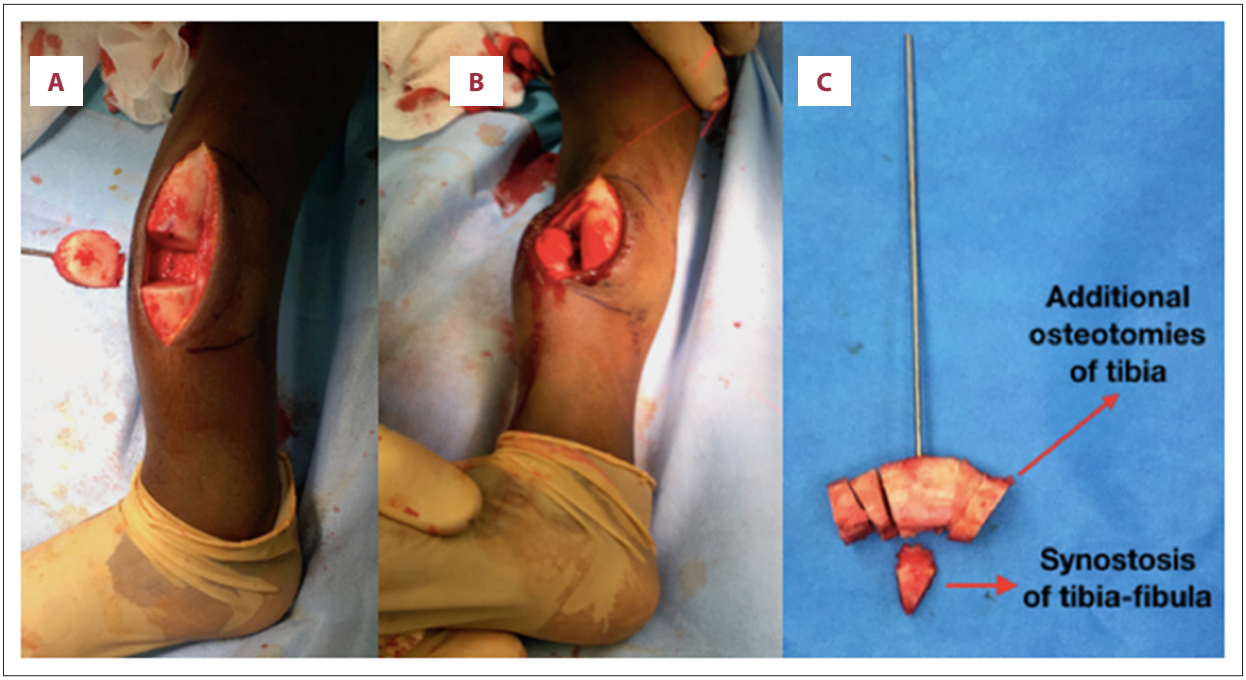


**Figure 6.** (**A**) The primary osteotomy was performed with an osteotome after drilling multiple holes. (**B**) Alignment of the bone-ends after the main apex of the deformity was excised. (**C**) To achieve good alignment, additional small osteotomies were needed, on the proximal and distal side of the tibia.

Ender nails, elastic nailing, or telescoping rods are preferred over plating [3,5,10,16]. Enright et al. report a high complication rate in children with OI treated with plating (69.2% complication rate) [16].

Regarding the intramedullary rods, these can be fixed or elongated rods with the advantage of elongated rods being that allows the longitudinal bone growth, but the diameter has to be small enough to not affect the physis [17]. Sterian et al. report in their publication that although with telescoping rods a long-lasting osteosynthesis can be obtained, arthrotomies and nail insertion through the joint cartilage is needed, leading in potential joint stiffness [18]. They concluded that Fassier Duval telescoping nailing is a good alternative that avoids these problems [18].

Sangasoongsong et al. support the view of using humeral nails in femoral fixation in patients with OI over Rush nails as they have a smaller diameter and provide the interlocking property which is better for rotational stability [19]. Mulpuri and Joseph report the results of a 10-year period of intramedullary rodding in 16 patients. They found that the post-operative fracture rate in the elongating rod group was 0.04 per person while in the non-elongating implant group the post-operative fracture rate was 0.21 per person [20].

Possible complications with intramedullary nailing can be bending of the rod, migration, disengagement, fracture of the rod or fracture of the bone, nonunion or delayed union, and hardware loosening [3,16,21]. Chiarello et al. analyzed 29 patients (245 procedures) and compared conservative treatment (e.g. casts) and surgical treatment (e.g., pinning, intramedullary nailing, plating) and although they found no significant difference regarding the complications between the 2 groups, they concluded that in type III OI the use of intramedullary devices in association with bisphosphonates appeared to be better [21].

Bisphosphonates have been extensively used for patients, with OI especially in children aged 3 years-old and they can be administrated for up to 2 years, but orthopedic surgery has the primary role in severe cases [14]. Georgescu et al. published the evidence from 32 operated patients with OI (81 surgeries) either with Sheffield telescoping rod, circular external fixator, or bone transplantation and they conclude that surgical treatment of moderate to severe long bone deformities was the only chance for these patients to walk again [22].

Pre-operative planning and selection of the best implant for patients with OI is very important but problematic as well. The age of the patient, the distortion of the anatomy, the advantages and disadvantages of the various surgical instruments, the availability of the different surgical instruments, the surgical experience, and the post-operative complications are all factors that should be carefully examined in order to select the best implant for each case [18,19]. With our case report, we present a relatively simple surgical technique for



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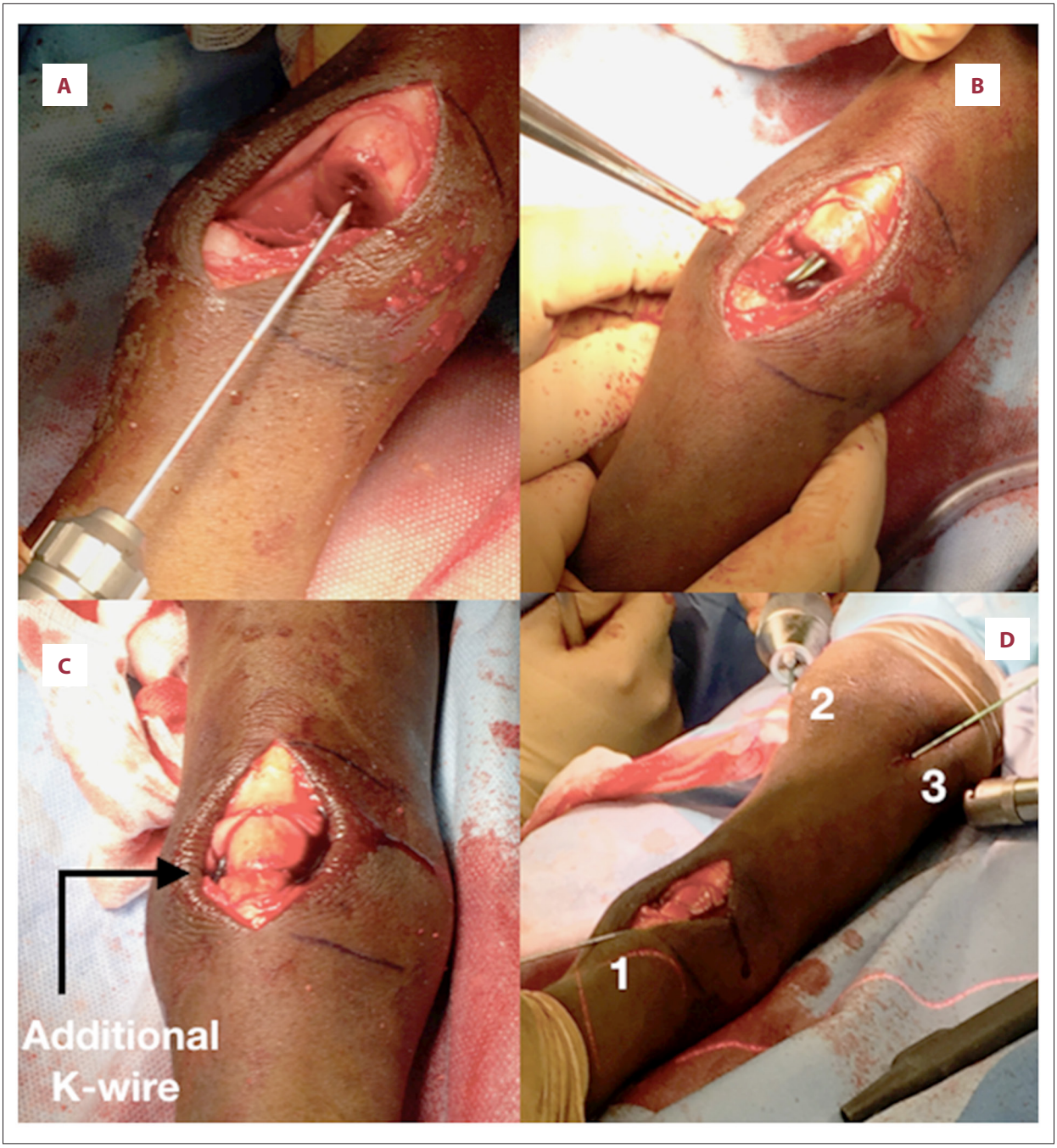


**Figure 7.** (**A**) Multiple drilling of the medullary canal in order to increase its diameter (due to the narrowing at this level). (**B**) After increasing the diameter of the medullary canal, forward of the pre-inserted nails to the distal part of the tibia. (**C**) An additional, third K-wire is inserted obliquely from the distal part to the proximal part. (**D**) Intraoperative image showing the stabilization of tibia after osteotomies and alignment. Number 1 indicates the additional extra-medullary K-wire, whereas numbers 2 and 3 indicate the intramedullary, initially inserted TENS nails. TENS – Titanium Elastic Nail System; K-wire – Kirschner wire.



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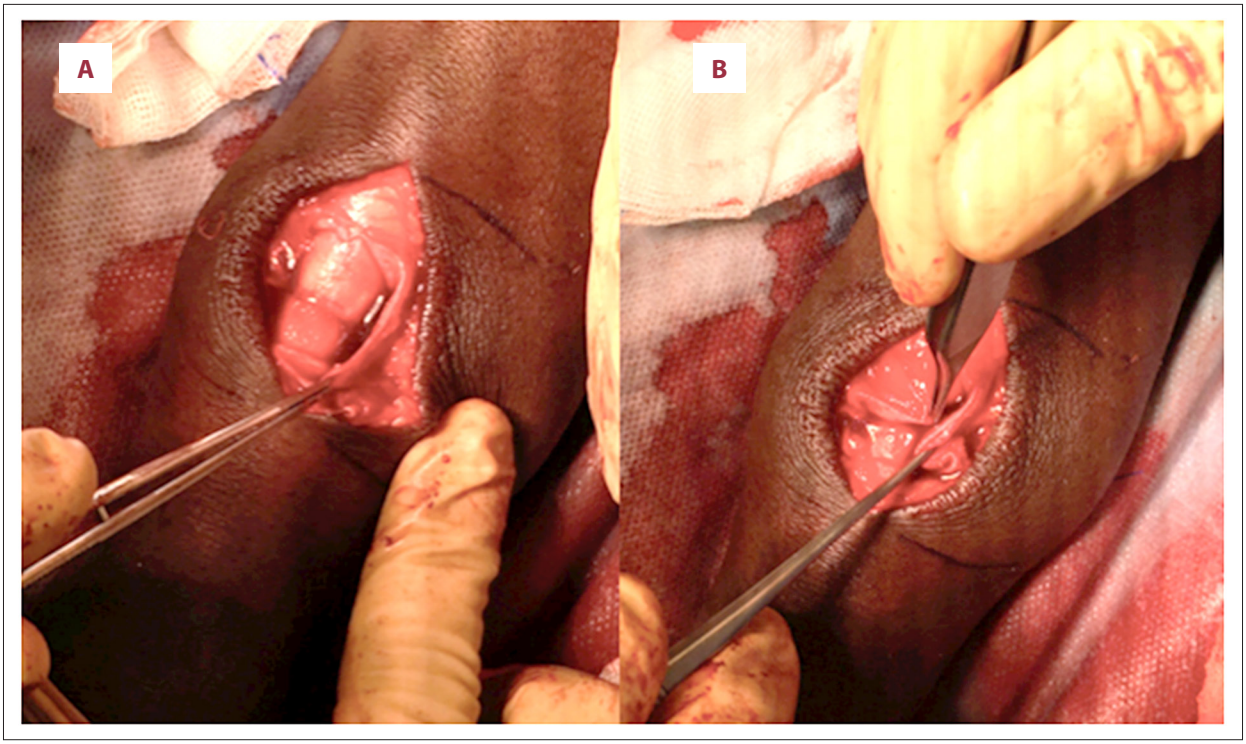


**Figure 8.** (**A, B**). After final alignment and stabilization of the performed osteotomies, an adaptation of the periosteum will be needed, and it is important to provide nutritional support to the bone.

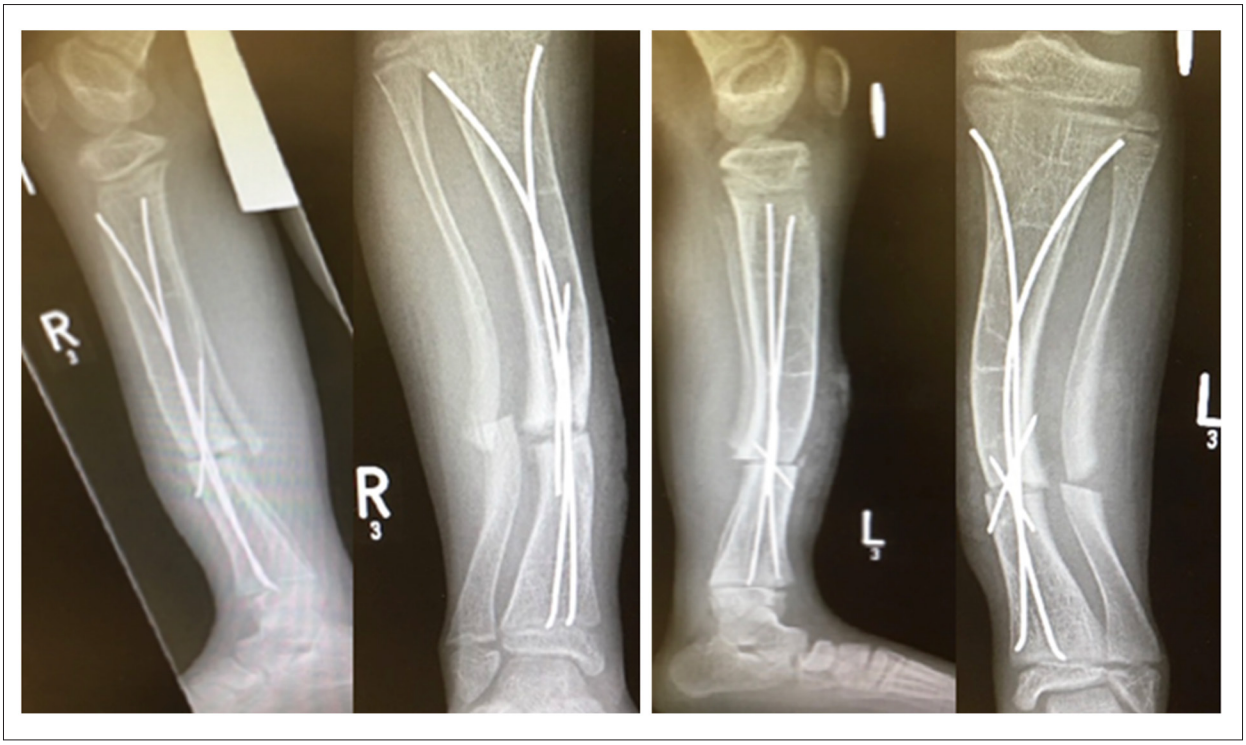


**Figure 9.** Post-operative f+profile x-ray views of the left and the right tibia, showing the correction of the extreme bowing deformity of the tibias.



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**Figure 10.** (**A, B**). Clinical presentation of our patient 15 days after surgery. No bowing deformity of the tibia can be observed.

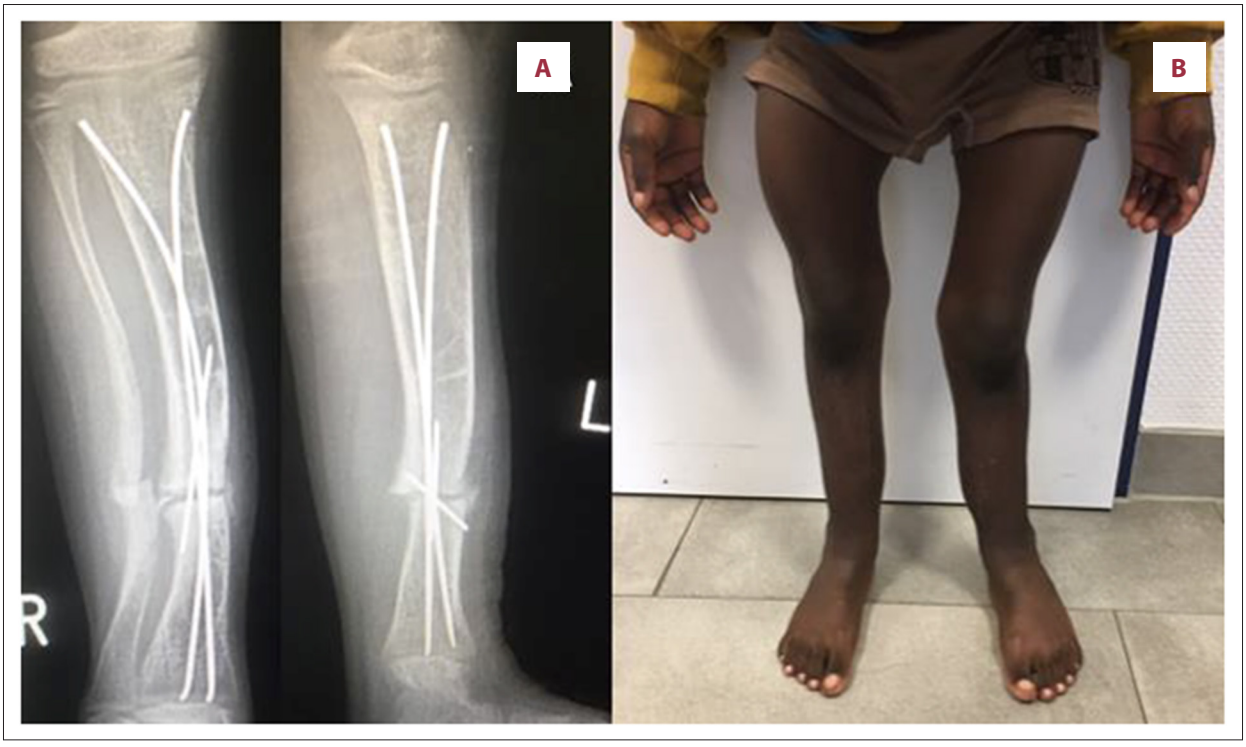


**Figure 11.** (**A**) X-ray of right tibia (f) and left tibia (profile) 6 months after surgery indicating that callus formation begins. (**B**) 1 year after surgery, our patient can stand and walk independently.



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correction of long bone deformities that can be a good alternative when there is no access to advanced, innovative surgical systems, such as telescoping rods.

## Conclusions

Our opinion is that each patient with OI is a unique case and the corrective surgical treatment should be adapted to each case. In patients with OI diagnosis, a multidisciplinary team approach by orthopedic surgeons, physicians, pediatricians, and endocrinologists should be performed. From the orthopedic point of view, the final goal should be the ability to walk independently with as minimal complications as possible. This goal can be achieved, regardless of which surgical technique is performed.

### Conflicts of interest

None.

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