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Straightening the Bow: Acute Bilateral Femoral Deformity Correction in an Adolescent with X-Linked Hypophosphatemic Rickets – A Case Report with 3 Years Follow-up

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Authors' contributions

This work was carried out in collaboration among all authors. Author MA designed the study and manuscript writing and literature review. Author GRD managed the analyses of the study. Other authors managed the literature research and part of operating team. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Introduction: X-Linked Hypophosphatemic Rickets (XLHR) occurs due to mutations in *PHEX* gene leading to a disordered production of FGF23 and hypophosphatemia. This disease is characterized by bowing of lower extremities. Phosphate supplements and oral vitamin medications, partially or, in some cases, fully restore the straightness of the limbs. Surgery is considered a vital alternative for severe or residual limb deformities in adolescent population.
Case Report: A 19 year old male presented to us with complaints of bilateral bowing of lower extremities, diagnosed to be X-Linked Hypophosphatemic Rickets .He underwent correction of deformity in both femurs, spaced 5 months apart, with intramedullary nailing, based on the principle of CORA. Follow-up period of 3 years showed a good functional outcome from the patient with no complications acquired so far. Patient gained a height of 5 cms.
Discussion: The treatment for XLHR, principally, starts with rectifying hypophosphatemia and, is

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achieved by vitamin D and phosphate supplementation prior to surgery. A variety of techniques are used in the treatment, of which acute correction and fixation with intramedullary nail, is considered to be superior to osteotomy or Ilizarov or plate fixation, as both rotation and angulation deformities can be corrected.

Conclusion: Surgery becomes the best choice of management at this stage. Deformities of the diaphysis are triumphantly treated with multiple osteotomies over an intramedullary rod at any age, preferably after puberty to prevent recurrence. It allows patients to approach their normal activities. A good result was achieved by using multiple osteotomies by CORA and intramedullary nailing.

Keywords: XLHR; deformity correction; rickets.

1. INTRODUCTION

"X-Linked Hypophosphatemic Rickets" refers to a group of disorders with biochemical and clinical features that induce multiplanar deformities of the lower limbs. It manifests usually around childhood leading to a delay in mineralization of growth cartilage and deformity of the bones around the knee. Biochemically, it is characterized by low serum calcium and phosphate levels [1,2]. Being the most common form of heritable rickets, it is caused by mutations impairing the function of the endopeptidase encoded by the *PHEX* gene, located on the X chromosome (Xp 22.1). Some individuals manifest features of XLHR with no family history of rickets (sporadic cases), and many subsequently transmit the phenotype in an X-linked dominant manner consistent with HPR. When PHEX is impaired, fibroblast growth factor 23 (FGF23) is produced in excess, leading to hyperphosphaturia and insufficient production of 1,25(OH)2 vitamin D, thereby altering the function of target organs, namely the growth plate and bone, causing delayed enchondral ossification and reduced growth rate . Children affected by XLHR present with rickets, osteomalacia, enlarged joints, enthesopathy, bone pain, growth retardation, abnormal mineralization of the teeth, dental caries and leg bowing [2]. XLH- rickets is characterized by complex deformities of lower limb including coxa vara, genu varum / valgus and tibial/ femoral bowing [1,3], shrinking the quality of life in children. In the case of chronic phosphaturia, there is a progression to osteoarthritis due to long term weight bearing on misaligned hips, knees and ankles [4], with burdensome day to day activities. Often misdiagnosed as physiological rickets or drug resistant forms, a family history of short stature points to XLHR [5]. Despite adequate medical management, many patients continue to have significant lower limb deformities and need operative management for prevention of secondary problems and cosmetic reasons. Surgical management is usually reserved for treating severe deformities. Here we present a case of a 19 year old male with bilateral bowing of the femur, both treated with Intramedullary nailing, spaced five months apart and correction of deformity, with a 3 year follow-up.

2. CASE REPORT

A 19 year old male presented to us with complaints of bilateral bowing of lower extremities, present from the age of 3 years. The deformity was of insidious onset, and progressed gradually to attain the present condition. It was not associated with pain and there was no history of trauma or any other constitutional symptoms. He experienced difficulty in walking and sitting cross-legged, affecting his day to day activities. Noticing his deformity at the age of three, his parents consulted a physician, and it was subsequently diagnosed to be rickets, for which he was administered Vitamin –D supplementation along with oral phosphate substitution for a long period. The patient presented to us after 15 years with the deformity, and the present diagnosis was initially missed but later clinched based on corroborative laboratory findings of hypophosphatemia , low borderline serum calcium (7.7mg/dl) and vitamin D Levels, and elevated alkaline phosphatase levels. A phosphate concentration of 1.6 mg/dL was present indicating significant lower levels. ALP being 288 IU/L.

On examination of bilateral lower limbs – bowing was present over bilateral femurs with no obvious deformity over leg segment [Fig. 1]. No diffuse or localized tenderness was present over the thigh segment. No warmth or dilated veins, with no thickening or broadening present. Range of movement of hip and knee had no restrictions and was not associated with any pain or muscle spasm. There was no distal neurovascular deficit or limb length discrepancy. Radiograph of pelvis

with both hips was done [Fig 2a] with bilateral lower limb scannogram, [Fig. 2b] which revealed bilateral bowing of femur with no obvious deformity of the tibia. The diagnosis was confirmed with patient having excessive phosphaturia. Consequent to failure of medical management, although early diagnosis and medical therapy yield better outcomes, we planned for osteotomy and internal fixation to create normal mechanical and anatomical axes as bone deformity in XLH consists of deviation of normal alignment of lower limb . Following the Paley principles of deformity correction, the center of rotation and angulation (CORA) was determined clinically and from radiograph. The CORA [Fig. 3] was drawn by taking the anatomical axes of the proximal femur and distal femur and their intersection points. The osteotomy site passes through the bisector line. The patient's femur has two CORA points. The patient was accordingly planned for surgery after obtaining patient's consent and anesthesia fitness.

Fig. 1. Pre- operative clinical picture of the patient showing his deformity in the femur

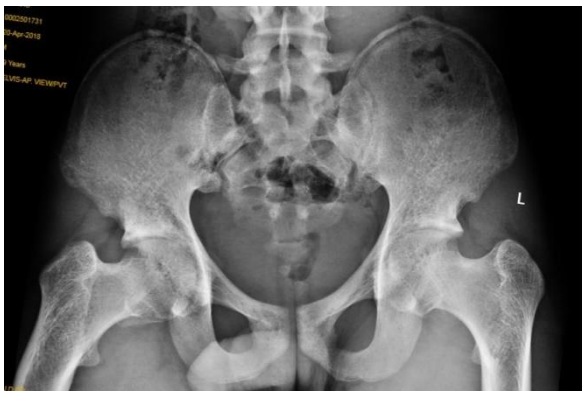

Fig. 2a. Preoperative pelvis radiograph

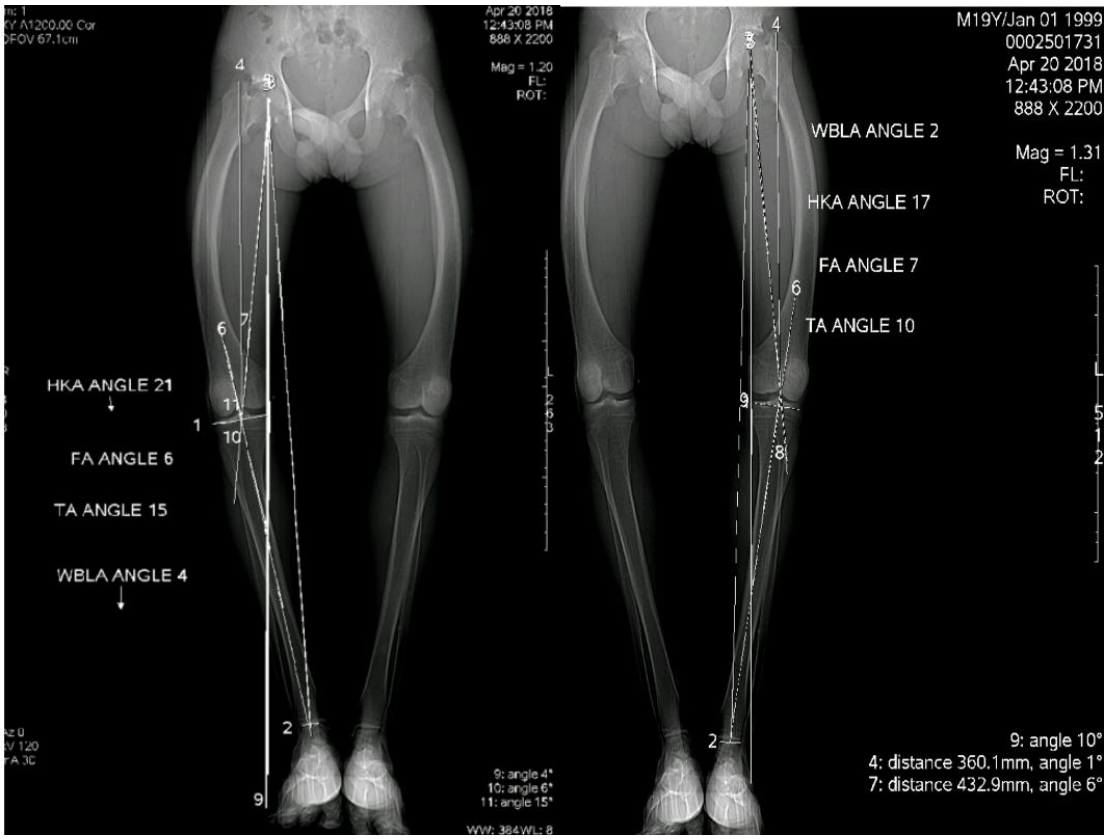

Fig. 2b. Preoperative CT scannogram of bilateral lower limbs

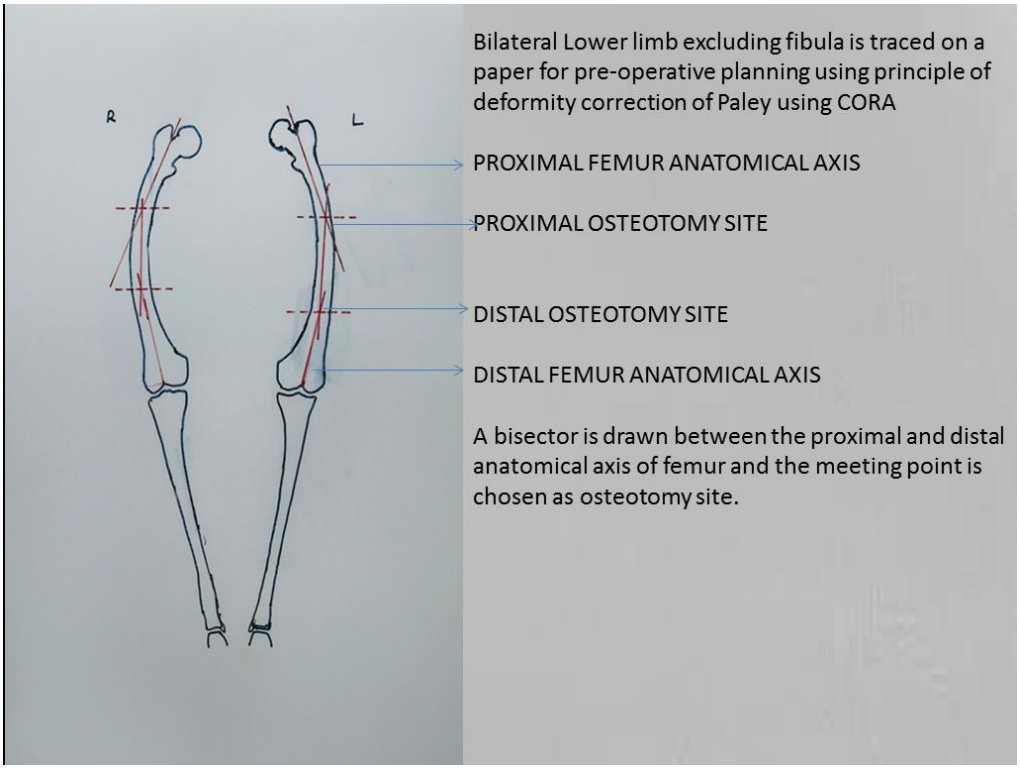

Fig. 3. CORA traced in a paper

Under GA, the patient was put in lateral position and two lateral incisions at the site of the proposed osteotomies were made initially. Two bone wedges were excised with a powered saw and removed to allow appropriate angular correction of the femoral shaft. The shaft fragments were realigned and a guidewire was passed from the proximal entry point across the greater trochanter into the distal femoral epiphysis. Following osteotomy the femur was internally fixed with adult femur nail of size 8.5 x 320mm (diameter 8.5mm, length 320mm) and with proximal and distal screws. The deformity correction and fixation of the femur was first done in right side [Fig. 4a, Fig. 4b]. Five months later the procedure was performed on the left femur [Fig. 5]. The aim was to correct the severe angulation of the left femur and to achieve the same length in both legs, The technique of multiple osteotomies and realignment by intramedullary fixation was performed in a similar way to the first operation using an adult femur intramedullary nail of the same size (8.5 x 320mm). The patient was allowed to mobilize as previously and no medical treatment was given. The patient was kept on Non weight bearing mobilization with walker support for 6 weeks followed by Partial weight bearing mobilization for another 2 months, converting to full weight bearing walking at 14 weeks post surgery. The same protocol was followed for the left femur starting on Non weight bearing mobilization with walker support for 6 weeks, converting to partial weight bearing for 8 weeks, following which he was started on full weight bearing at almost 14 weeks post surgery. Post operatively patient was followed up at regular intervals of 6 weeks and then 3 months interval, 1 year [Fig. 6a, 6b] upto 3 years post surgery [Fig. 7a, 7b, 7c]. Radiologically union was achieved at the end of 8 weeks. A gain of 5 cm was achieved postoperatively following bilateral femur deformity correction. At the end of 3 years, no complications were expected in the patient with full knee ROM and patient is mobilizing well without any aids, having improved his quality of living. Follow - up radiograph showed corrected femoral bowing with healing of the osteotomies. Removal of implant has not been planned so far.

3. DISCUSSION

Deformities of lower limbs in adolescents with XHPR are markedly seen by their clinical appearance, laboratory data and biochemical analysis. The treatment for XLHR, principally, starts with rectifying hypophosphatemia and, is achieved by vitamin D and phosphate supplementation prior to surgery [1].

Although patients are reported to benefit from pharmacological supplements, progression to deformity is seen nevertheless [1].

The risk of complications is higher in cases operated at a younger age, and in order to reduce the rate of surgeries, a multidisciplinary approach can be undertaken with pharmacological treatment [2].

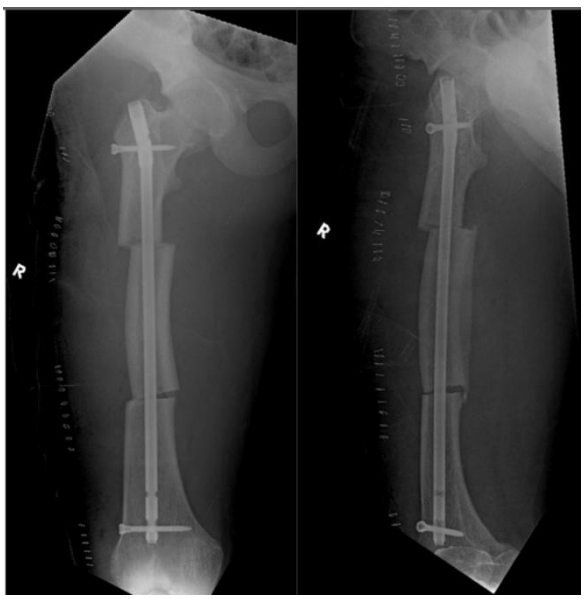

Fig. 4a. Immediate postoperative radiograph after Right femur deformity correction

Fig. 4b. Clinical picture after deformity correction of right femur

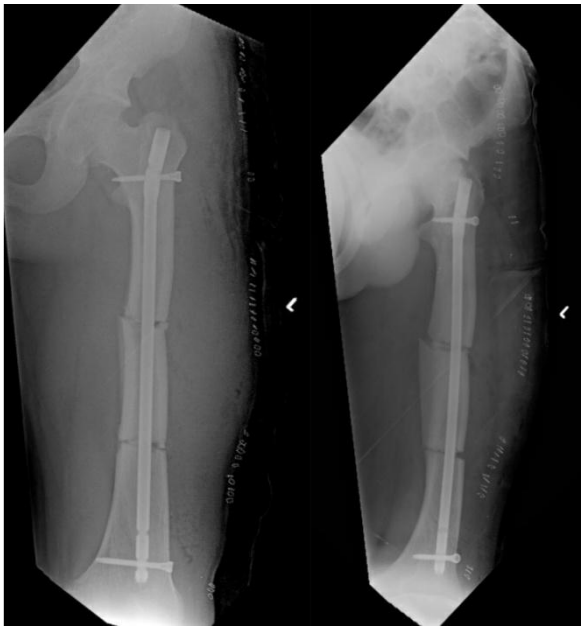

Fig. 5. Postoperative radiograph after left Femur deformity correction

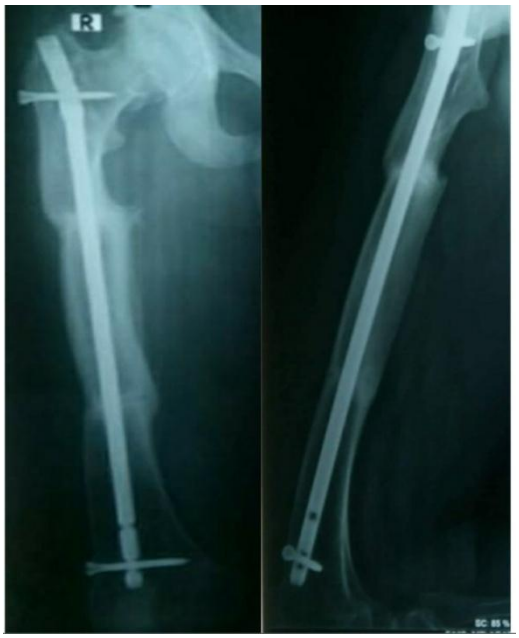

Fig. 6a. 1 year postoperative radiograph after right femur deformity correction

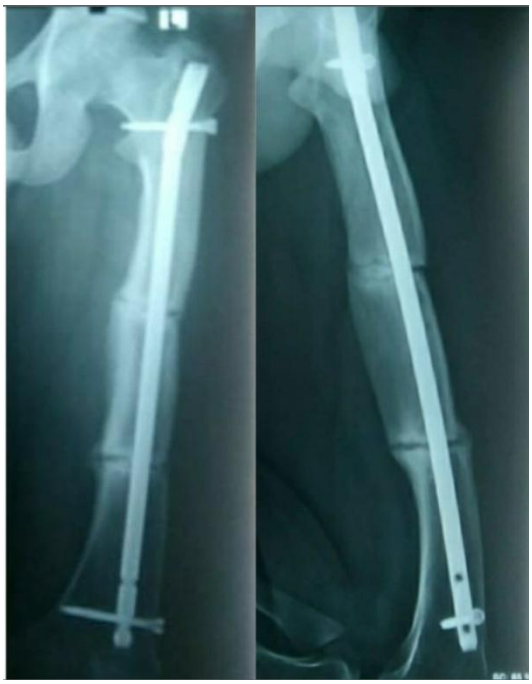

Fig. 6b. 1 year postoperative radiograph after left femur deformity correction

A variety of techniques for treating such a deformity includes acute correction and fixation of osteotomies using plates, external fixators, Taylor frame and intramedullary nailing with or without bony lengthening. A number of authors agree that severe deformities of the femoral shaft are most successfully treated by the technique of multiple osteotomies and fixation with an intramedullary nail. The first to report this, in 1959, Sofield and Millar [6], performed the technique of fragmentation and realignment by intramedullary fixation for the correction of deformities of long bones in children (n = 52) with congenital pseudarthrosis, osteogenesis imperfecta or fibrous dysplasia for limb lengthening. Intramedullary nailing can halt the deformity of diaphysis [3].

Fig. 7a. Clinical Picture after 3 years since the deformity correction of bilateral femur

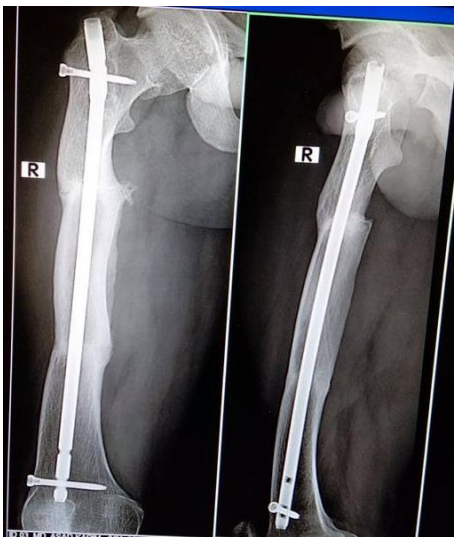

Fig. 7b. 3 year postoperative radiograph of right femur deformity correction

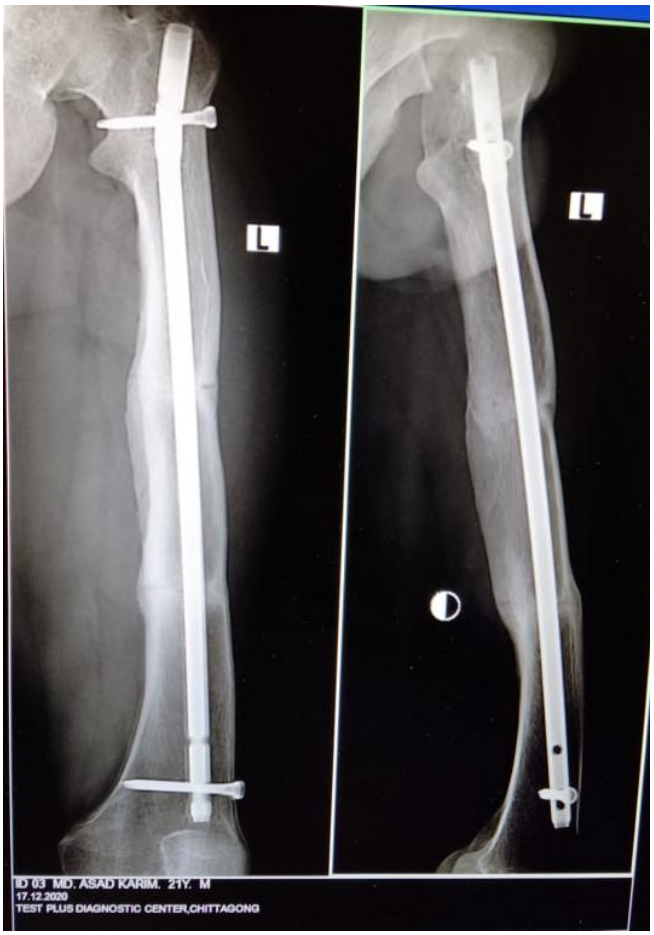

Fig. 7c. 3 year postoperative radiograph of left femur deformity correction

In our case report, the surgery was done at 19 years of age and it is always profitable to intervene at later stages when growth halts favouring only one corrective surgery after obtaining metabolic control over the disease to prevent recurrence and non-union [2,7]. However this statement does not strictly hold good for all cases, wherein some patients may require an early operation in major deformities that induce joint complications such as femoro-tibial instability, or patella sub-dislocation, or if the joints are at risk. Additionally long term administration of calcium and oral phosphate is associated with the risk of nephrocalcinosis, hypercalciuria and hyperparathyroidism posing a threat to children. The focus of surgical treatment should be a well aligned neutral lower limb and mobile pain free relaxed joints [7].

In our case, the patient did not benefit from medical therapy and deformity correction with intramedullary nailing became essential. As stated earlier, Sofield and Miller [6] held intramedullary nail deformity correction to be superior to osteotomy or Ilizarov or plate fixation as both rotation and angulation deformities can be corrected but not near joint line. Plate fixation proved difficult for angulation and rotation improvement. Furthermore if there is vascular compromise to the lower limb immediately after surgical lengthening, then immediate derotation of the bone is easier with intramedullary nailing than plates [8].

In bowed legs, irrespective of whether the CORA was in the femur or tibia, the knees are affected more by lateral joint laxity than medial [8], and so a gentle overcorrection in genu valgum, will shift the mechanical axis to medial knee compartment, giving a steady ligament construct. The CORA in our case was based on Paley et al [9], and even though it was not restored completely, a more favorable outcome was obtained by the patient at the end of 4 years. In a study done by Garcia-Cimberlo et al, the complication rate in using a femoral nail was very low with no recurrence of the deformity at further follow-ups.

The disadvantage is that, intramedullary nailing cannot allow early weight bearing due to its limited stability [3] and does not correct the articular inclination [10]. The selection of the intramedullary nail is based on surgeon's preference and patient specific. Skeletally mature patients may benefit from the placement of intramedullary rods for osteotomy fixation as they can span the bone and provide long term support.

4. CONCLUSION

The treatment of XLH rickets commences with medical therapy, which if diagnosed and started early, may aid in spontaneous regression of lower limb deformities. A delay will lead to angular and rotational deformities. Surgery becomes the best choice of management at this stage.

Deformities of the diaphysis are triumphantly treated with multiple osteotomies over an intramedullary rod at any age, preferably after puberty to prevent recurrence. It allows patients to approach their normal activities. A good result was achieved by using multiple osteotomies by CORA and intramedullary nailing and hence we prefer to use this procedure for similar cases of deformity correction. This case is of interest as neglected medical treatment has led to a deformity which now has a good mid-term follow-up

CONSENT

Consent has been obtained from the patient promising to avoid disclosure of personal details.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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