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Original Research Paper

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METABOLIC AND ORTHOPEDIC ASPECT OF X-LINKED VITAMIN D-RESISTANT HYPOPHOSPHATEMIC RICKETS

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ABSTRACT

INTRODUCTION: - The X-linked vitamin D-resistant hypophosphatemic rickets (VDXLR) is a metabolic disorder. Medication treatment consists of oral phosphate substitution and supplementation of active vitamin D compounds. Our study aimed to review our patients with VDXLR, focusing on those undergoing surgery, mainly lengthening procedures. The main parameters of interest were growth, height, the axis of the lower limbs, pain, and degenerative arthropathy.

METHODS: - Twelve patients with VDXLR were followed at our institution. Eight patients underwent surgical correction, and three of them in combination with bone lengthening. The corrections were executed at the end of growth in the patients. Clinical end points were height, leg axis, and pain.

RESULTS: - Single bilateral surgical correction was performed in six patients; one patient had three and five corrections. Bone lengthening was performed in three patients. At the last follow-up, the height of seven operated patients was within normal range. In addition, the leg axis was normalized in six patients with mild genua vara in two. Bone healing was excellent, and no surgical complications. There was no one radiological evidence of degenerative arthropathy.

CONCLUSIONS: - In case of bone deformity, surgery can safely be performed, independent of age or bone maturation. All patients were happy with the outcomes of axial corrective surgery and bone lengthening, and in the majority. Only one corrective intervention was needed.

KEYWORDS : Lengthening, Growing children, Rickets

INTRODUCTION: -

The X-linked vitamin D-resistant hypophosphatemic rickets (VDXLR) is a metabolic disorder first described by Albright et al.[1] Clinical features, presenting with the beginning of walking, include short stature, reduced growth rate, and bone deformities, such as coxa vara, femoral and crural bowing, and genua vara and valga . **(Fig- 1.a,b)** Medication treatment consists of oral phosphate substitution and supplementation of active vitamin D compounds.[2] Despite correct substitution, the main symptoms, i.e., deformity, pain, small stature, and—in some cases—also fractures, can occur. The choice of implant depended on the bone maturity of the patient and the surgeons' preference. Most authors proposed surgery at or close to bone maturity because of the risk of recurrence of bone deformity and non-union in growing bones.[3,4] They also reported complications such as recurrent deformity, joint stiffness, deep intramedullary infection, refracture, nerve palsy, leg length discrepancy, and pin tract infection. The main parameters of interest were growth, height, the axis of the lower limbs, pain, and degenerative arthropathy.

Figure-1

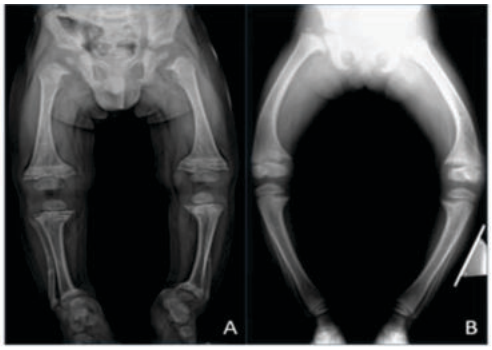

Fig-1 A.B Severe radiological deformities of the lower extremities of pt. 1 at his first consultation

MATERIALS AND METHODS: -

This study was performed from Jan 2019 to 2020 in the Department of orthopedics, IGIMS Patna-14 Bihar. Twelve patients were included with VDXLR in this study. Four patients were only on medical treatment, and eight patients underwent surgery, including three with lengthening procedures. Radiological evidence of rickets, lower limb deformity, or swollen metaphysis of the wrist and ankle in the physical examination was not mandatory. All patients received daily oral phosphate (45–100 mg/ kg, maximal dose 2 g) and calcitriol (0.01–0.05 lg/kg). Pain and drug compliance were recorded. Plain X-rays were only ordered in case of florid rickets or suspected degenerative joint alteration. Bone maturity was defined according to bone age (X-ray of hand and wrist).

RESULTS AND DISCUSSION: -

We followed 19 female and three male patients diagnosed with VDXLR. Eight patients underwent surgery. Six patients had a single bilateral surgical correction, whereas one patient each had three and five operations. Three patients underwent bone lengthening using an external fixator system with a lengthening rate of 1 mm per day. At last follow-up, five patients had a mature skeleton, and seven, including four, treated surgically, were still growing. Six patients had single bilateral corrective surgery. The underlying deformity was genua vara, crura vara, and genua valga in three, two, and one patients. The deformation for the genua vara ranged from 12 to 33°, crura vara from 14 to 19°, and genua valga from 14 to 25°. An infracondylar valgization corrected genua vara with a K-wire fixation in one patient and plates in two patients. Crura vara was corrected with corticotomy of the tibia and fibula and simultaneous bone lengthening in two patients. The patient with genua valga was corrected by a bilateral supracondylar variation using plates. One patient had five bilateral corrective interventions, two corrective manipulations using the external fixator in situ, and twice an osteosynthesis of a fracture with refracture of the right femur. This boy had presented as an asylum seeker at the age of 9 years with very short stature (well below the third centile), pain, and multiple deformities, such as coxa vara, femora vara, and crura

antecurvata **(Fig- 1a,b).** The first intervention included acute extension osteotomies of the tibiae and corticotomies of the fibulae; the tibiae were fixed with compression screws. One year later, a subtrochanteric validation was performed and fixed with an external fixator. This step was staged with a time delay of 3 months. A femoral lengthening by 33 mm was performed. At the age of 11 years, a corticotomy with bone lengthening of both tibiae and fibulae (35 mm) was performed with correction of crura vara. At 12 years, a bilateral femoral shaft corticotomy was performed with an additional elongation of 50 mm. Six months after consolidation, a right femoral shaft fracture occurred. An elastic nailing procedure was performed with uneventful consolidation. Because of femoral varus deformity, the last corrective surgical intervention was a bilateral femoral shaft corticotomy on two levels at the age of 15 and three months using an intramedullary nail. At least follow-up, he was pain-free, and his height was in the third centile (165 cm). **(Figure-2)** Vitamin D-resistant hypophosphatemic rickets (VDXLR) is a rare metabolic disease that needs close cooperation between disciplines. Eight patients had corrective osteotomies for axial deviation, three of them also a lengthening procedure. In the majority of patients, a single bilateral simultaneous intervention restored the correct axis. The study of Dudkiewicz concluded that though a metabolic bone disease is not a common indication for lengthening and could be problematic, lengthening can be considered an option.[5] This combination makes sense to prevent refracture, but the complication of deep intramedullary infection is very severe and has to be evaluated with the risk of refracture. Our complications with lengthening were minor. Joint stiffness resolved after stopping lengthening. Pin tract infection healed after antibiotic therapy. We agree with Dudkiewicz's report and the study of Song et al.[6] The results of our small group demonstrate that the lengthening technique is a good option for short stature also in compliant patients with metabolic bone disease.

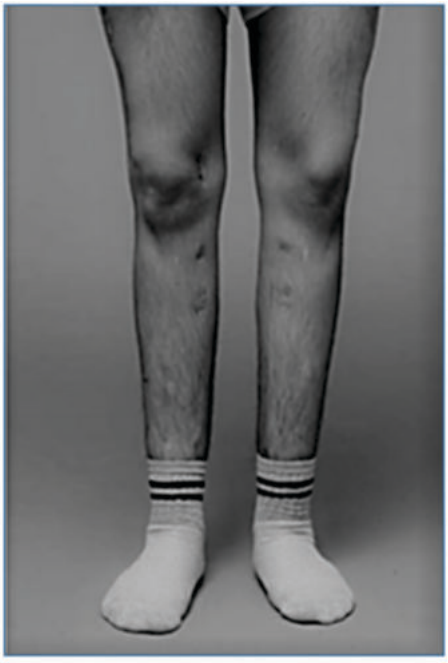

Fig. 2 Lower extremities of patient 1 at his last consultation with correct axes

CONCLUSIONS: -

In case of bone deformity, surgery can safely be performed, independent of age or bone maturation. All patients were happy with the outcomes of axial corrective surgery and bone lengthening, and in the majority. Only one corrective intervention was needed. The role of the lengthening procedure to improve short stature needs further studies. Close cooperation between pediatricians and orthopaedic surgeons is mandatory for optimal care of these patients.

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