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Case report: Epiphysiodesis for knee deformity correction in hypophosphatemic rickets

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Abstract

Introduction: Vitamin D-resistant rickets, or hereditary hypophosphatemic rickets, encompasses a group of disorders in which normal dietary intake of vitamin D is insufficient to achieve normal mineralization of bone because of pathologic renal phosphate wasting. X-linked dominant disease involves (PHEX) gene mutation, which produces elevated levels of FGF23 that reduces renal phosphate reabsorption and conversion of 25(OH) calciferol to its active form, 1, 25 (OH) 2calciferol. This leads to increased renal phosphate excretion, hypophosphatemia, short stature, long bone bowing, physeal widening and rachitic rosary. In autosomal dominant form, mutations in FGF23 produce renal phosphate wasting. In the autosomal recessive form, mutations in DMP1 gene and ENPP1 impair osteocyte maturation and bone mineralization, producing elevated levels of FGF23, leading to phosphaturia and hypophosphatemia.

Methods and Materials: Case was of 16 yrs old male with Right Genu Valgum, Left Genu Varum with short stature in k/c/o medullary sponge kidney. Patient developed windswept deformity in both knees since age of 14 years. Xray of both Knee showed angle of 17/163 on Right and 165/15 on Left. Blood investigations showed normal levels of calcium (8.6), PTH (59.7). Serum phosphate concentration was significantly decreased (2.5), whereas the 1, 25 (OH)-cholecalciferol level was low in response to hypophosphatemia. Serum ALP concentration was elevated (400). Urine showed increased phosphate in urine. Medical treatment comprised of oral replacement of phosphorus and active form of vitamin D, calcitriol. In surgical treatment, case was selected for Open reduction-Internal fixation with Recon plating (Epiphysiodesis) in both knees.

Results: Xray/Scanogram of both knee joint done at Immediate postop showed 164/16 (Rt):165/15(Lt), at 6 months showed 169/11(Rt): 170/10(Lt) & at 9 months showed 172/8(Rt):174/6(Lt). Epiphysiodesis has resulted in correction at Right knee and at Left knee with 1 degree per month.

Conclusion: Epiphysiodesis restricts the growth naturally occurring at one side of physes and this facilitates growth at other side of physes. It helps in correction of deformity gradually, at around 1 degree per month as it depends on skeletal maturation. Epiphysiodesis is done before physeal closure and has no role after achieving skeletal maturity.

Keywords: Hypophosphatemic rickets, windswept knee deformity, scanogram, epiphysiodesis

Introduction

Vitamin D-resistant rickets, or hereditary hypophosphatemic rickets, encompasses a group of disorders in which normal dietary intake of vitamin D is insufficient to achieve normal mineralization of bone because of pathologic renal phosphate wasting.

X-linked dominant disease is the most frequent form of hereditary rickets, with an incidence of 1 in 20,000. (PHEX) gene mutation, which produces elevated levels of FGF23 that reduces renal phosphate reabsorption and the conversion of 25(OH) calciferol to its active form, 1,25 (OH)2calciferol. This prevents the normal compensatory increase in active vitamin D formation associated with hypophosphatemia. Thus leads to increased renal phosphate excretion, hypophosphatemia, short stature, long bone bowing, physeal widening and rachitic rosary.

In autosomal dominant form, mutations in FGF23 have been identified that produce renal phosphate wasting.

In the autosomal recessive form, mutations in DMP1 gene and ENPP1 impair osteocyte maturation and bone mineralization. And produce elevated levels of FGF23, leading to phosphaturia and hypophosphatemia.

Radiological investigations include Xray of both Knee joint- AP, lateral views and Scanogram of both lower limbs. Blood investigations include serum phosphate, 1, 25(OH)-cholecalciferol level both of which are decreased.

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Serum alkaline phosphatase (ALP) concentration are elevated whereas serum calcium and PTH tend to be normal. Urine shows increased phosphate levels.

The Medical treatment consists of oral replacement of phosphorus in large doses and the administration of an active form of vitamin D, calcitriol.

Epiphysiodesis is the surgical treatment of choice as it restricts the growth naturally occurring at one side of physes and this facilitates growth at other side of physes. In other surgical treatments, multilevel osteotomy is generally required to satisfactorily correct the mechanical axis of the limb. External fixation allows fine tuning of the alignment postoperatively, when the patient is able to stand. Others advocate the use of intramedullary fixation.

Methods and Materials

The case report comprised of a 16 yrs old male residing in Vyara, Gujarat, having – Right Genu Valgum, Left Genu Varum [Fig.2] [Fig.3] with short stature in k/c/o Medullary sponge kidney [Fig.1]. Patient had growth retardation since age of 1.5 years and then developed windswept deformity in both knees, difficulty in walking since age of 14 years and brought to New Civil Hospital, Surat.

Xray of both Knee joint AP, lateral views- showed angle of 163/17 on Right and 165/15 on Left.

Blood investigations showed normal levels of calcium (8.6), PTH (59.7). The serum phosphate concentration was significantly decreased (2.5), whereas the 1, 25 (OH)-

cholecalciferol level were low in response to the hypophosphatemia.

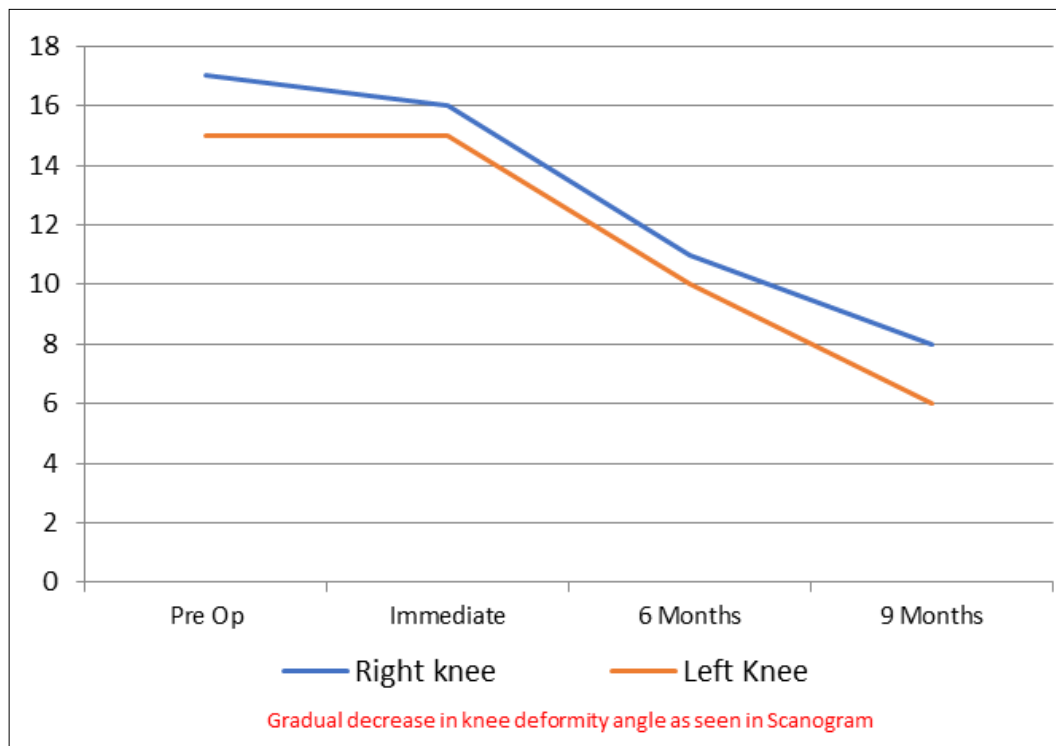
Serum alkaline phosphatase (ALP) concentration was elevated (400). Urine showed increased phosphate in urine.

Surgical Technique

The Case was selected for Open reduction+ Internal fixation with Recon plating (Epiphysiodesis) in both knees. For Left Genu Varum, Lateral Epiphysiodesis was done. 6-7 cm incision made over left knee lateral aspect and distal femur & proximal tibia exposed. 3 hole Recon plate placed over lateral aspect of distal femur and locked with two 3.5 locking cortical screw, each in metaphysis and epiphysis. Another 3 hole recon plate placed over lateral aspect of proximal tibia and locked similarly. For Right Genu Valgum, Medial Epiphysiodesis was done, 3 hole recon plate placed over medial aspect of distal femur and 3 hole recon plate placed over medial aspect of proximal tibia and locked similarly to opposite side

Results

Xray/Scanogram of both knee joint done at Immediate postop showed 164/16(Rt):165/15(Lt) [Fig.4]. At 6 months showed 169/11(Rt): 170/10(Lt), At 9 months showed 172/8(Rt):174/6(Lt) [Fig.5] [Fig.6]. Epiphysiodesis has resulted in correction at Right knee and at Left knee with 1 degree per month [Table 1].



Graph 1: Gradual decrease in Knee deformity at Preop, Immediate Postop, 6 month Postop & 9 month Postop at rate of 1 degree per month

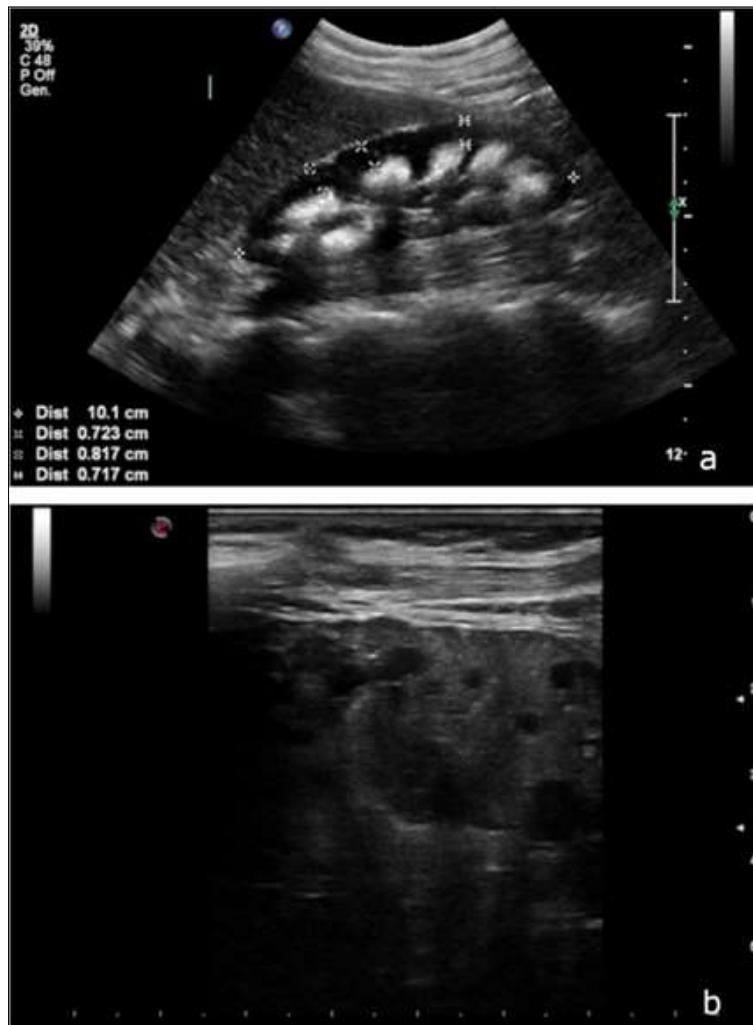


Fig 1: USG findings of Medullary sponge kidney in 16 year old Patient with Hypophosphatemic Rickets



Fig 2: Clinical picture of Right Genu Valgum & Left Genu Varum deformity at initial presentation.

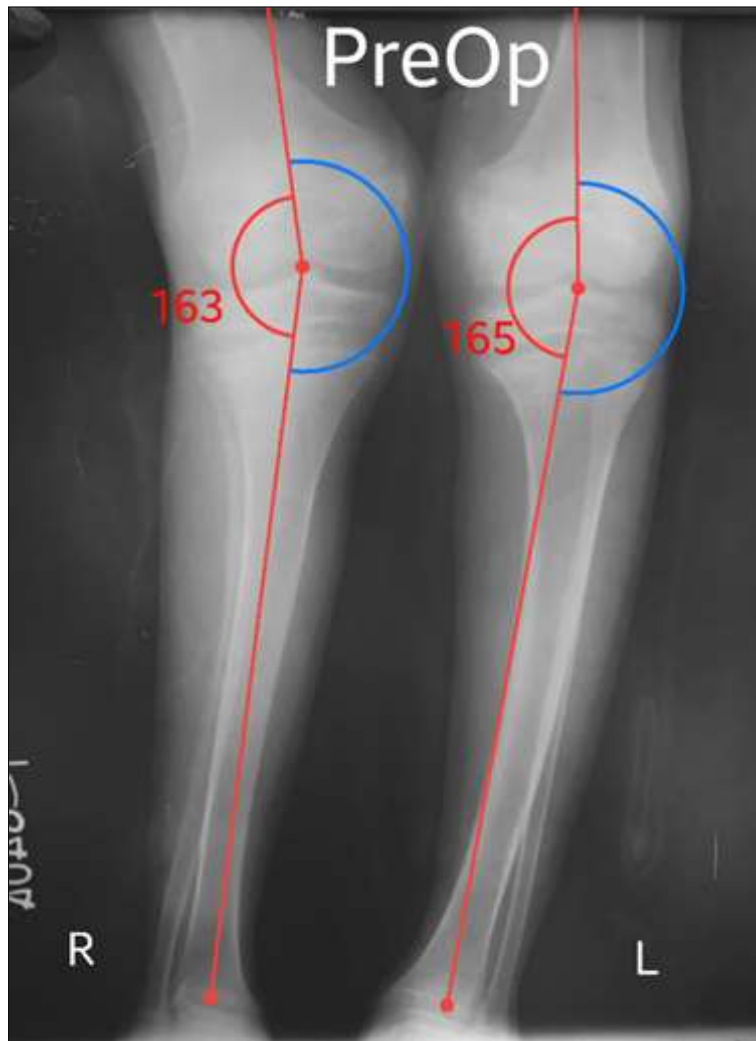


Fig 3: Preop AP Xray of both knee showing significant Windswept deformity (Right Genu Valgum & Left Genu Varum).

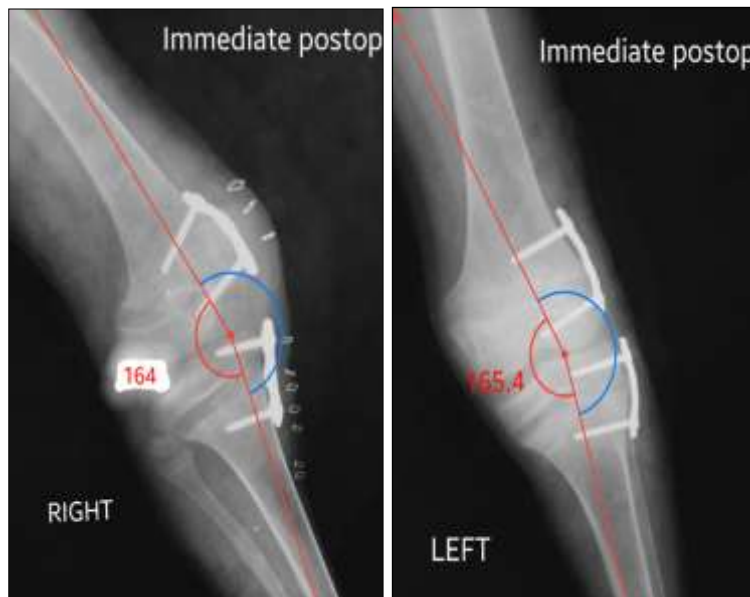


Fig 4: Immediate Postop AP Xray of both knee showing Medial Epiphysiodesis for Right Genu Valgum & Lateral Epiphysiodesis for Left Genu Varum.

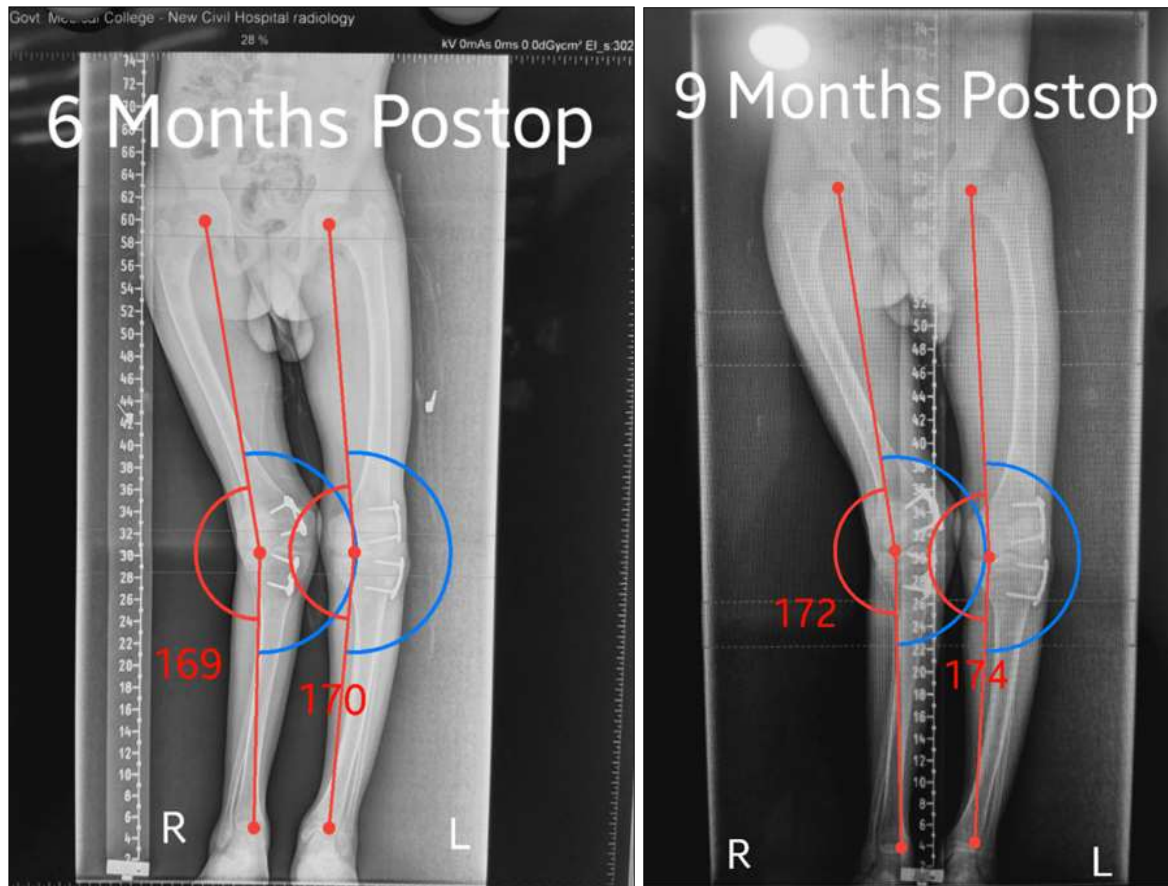


Fig 5: Postoperative Scanogram of bilateral Lower Limbs at 6 months (Left) & 9 months (Right).



Fig 6: Clinical picture at 9 months Postoperatively

Conclusion

Epiphysiodesis restricts the growth naturally occurring at one side of physes and this facilitates growth at other side of physes.

It helps in correction of deformity gradually, at around 1 degree per month as it depends on skeletal maturation. Epiphysiodesis is done before physeal closure. It has no role after achieving skeletal maturity. Other surgical treatments like Multilevel osteotomy, External fixation and Intramedullary fixation are more demanding and require meticulous follow up and are more prone to failure.

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