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# X linked Hypophosphatemicrickets (PHEX mutation), Badi Alenazi

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## Background

X-linked Hypophosphatemic rickets (XLHR) is a rare form of rickets that affect children. High index of suspicion is needed to diagnose this entity.

## Aim

To report a case of XLHR in a Saudi boy.

## Case Report

An 18 month old boy presented with delayed walking, lower limb deformity and persistent radiological rachitic changes. A diagnosis of nutritional rickets was made by general practitioner at 13 months old and was started on oral cholicalciferol 4000 IU once daily. There was no improvement on follow-up despite good compliance. Parents were not consanguineous and there was no family history of similar condition. On examination his growth was satisfactory but had waddling gait, frontal bossing. Bilateral Harrison's salcui and rachitic rosary. His serum calcium 2.4 mmol/l (2.1- 2.5) phosphorus 0.6 mmol/l (0.8- 1.5), Alkaline phosphatase 1050 IU/L (50 - 136), Mg 0.8 mmol/l (0.7 - 1.03). Parthyroid hormone 6.4 pmol/l (1.59-6.89), 25 OH vitamin D 214 nmol/L (50 - 250) and 1,25 (OH )2 vitamin D 68 pmol/L (38 - 133). He had normal serum urea and electrolytes, liver function tests and arterial blood gas. X- Ray knees and wrist showed cupping and fraying of the metaphysis. Direct sequencing of the PHEX gene confirmed the diagnosis of XLHR. He was treated with Alfacalciferol and oral phosphorus. His repeated bone profile improved but he still has significant bowing of the legs at the age of 3 years.

## Conclusion

XLHR should be considered as a cause of rickets even in areas with high prevalence of vitamin D deficiency like KSA.