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REVIEW OF HYPOPHOSPHATAEMIC RICKETS IN HOSPITAL PUTRAJAYA

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INTRODUCTION
Hypophosphataemic rickets (HR) is a rare cause of rickets characterized by renal phosphate wasting. X-linked dominant rickets (XLH) is the most common type of HR caused by inactivating mutations of PHEX.

METHODOLOGY
We reviewed medical records of 6 patients from the paediatric endocrine clinic in Hospital Putrajaya who presented with clinical and biochemical features of HR. DNA samples were sent for whole exome sequencing (WES) with the collaboration of Faculty of Medicine and Health Sciences, UPM.

RESULT
There were 2 male and 4 female patients from 5 families included in this review. The median age at onset and diagnosis was 1.75 years old (1-4 years) and 3.7 years old (1.8-5.9 years), respectively. The presenting features included bowed legs (100%), wrist swelling (100%), gait disturbance (50%) and limb pain (33%).

Laboratory findings at diagnosis included mean serum calcium and phosphate levels which were 2.34 ± 0.07 mmol/L and 0.72 ± 0.06 mmol/L, respectively. The mean alkaline phosphatase level was 591 ± 169 IU/L. The serum 25(OH)D_3 was 66.43 ± 24.3 nmol/L (normal: 50-250 nmol/L). The serum iPTH was 4.85 ± 2.5 pmol/L (normal: 1.3-9.3 pmol/L). Tubular reabsorption of phosphate was available for 2 patients which were 72 and 77% (normal: >85%). All patients had normal urine calcium/creatine ratio for age. XLH was confirmed by WES in 4 patients from 3 families. 2 patients had no pathogenic mutation detected. All the patients were treated with multi-dose oral phosphate and active vitamin D analogue. However, the majority developed progressive bowing of lower limbs, in which half of the patients required corrective osteotomy. At the latest follow-up, they had lower mean height SDS (-3.2 ± 0.63) and higher mean iPTH levels (13 ± 7.19) compared to their initial presentation.

CONCLUSION
XLH is the most common cause of HR in this hospital. The challenges associated with a multi-dose oral phosphate regimen include worsening height SDS and iPTH levels. New treatment approaches may help to mitigate these negative effects.