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HEALTH-RELATED QUALITY OF LIFE IN CHILDREN AND ADOLESCENTS WITH X-LINKED HYPOPHOSPHATEMIA (XLH) AT UNIVERSITI MALAYA MEDICAL CENTRE

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INTRODUCTION
X-linked hypophosphatemia (XLH) is a rare genetic disorder caused by PHEX mutations, characterized by chronic hypophosphatemia and renal phosphate wasting, resulting in skeletal, dental, and extra-skeletal complications. To date, no data from Malaysia are currently available on clinical characteristics or health-related quality of life (HRQoL) in paediatric patients.

METHODOLOGY
This cross-sectional study evaluated HRQoL among children and adolescents with XLH, using validated PROMIS (Patient-Reported Outcomes Measurement Information System) instruments, sociodemographic and clinical factors associated with HRQoL. This includes children and adolescents with confirmed XLH followed up at Universiti Malaya Medical Centre (UMMC) between November 2024 and March 2025.

RESULTS
Seventeen patients were analyzed (76.5% female and 23.5% male). The mean age at symptom onset was 2.82 ± 2.42 years, with a mean age at diagnosis of 5.21 ± 3.29 years. All patients continued to have musculoskeletal complications, including short stature (76.5%), bowing of legs (76.5%), bone/joint pain (47.1%), muscle pain (47.1%), and dental complications (52.9%). Serum alkaline phosphatase improved significantly ($p = 0.011$); persistent hypophosphatemia and ongoing musculoskeletal manifestations indicated suboptimal disease control. Elevated parathyroid hormone levels and an increase in urine calcium-to-creatinine ratio ($p = 0.020$) lead to secondary hyperparathyroidism and nephrocalcinosis. Adherence to conventional therapy was poor. Non-adherence was associated with worse pain outcomes (higher pain interference [$p = 0.037$] and greater pain intensity [$p = 0.025$]). PROMIS scores revealed severely impaired mobility (mean T-score 31.31 ± 12.17), increased fatigue (mean T-score 54.22 ± 7.60), and high pain interference (mean T-score 63.11 ± 9.50). Larger household size was also strongly associated with higher pain intensity ($p = 0.004$).

CONCLUSION
Malaysian children and adolescents with XLH continue to have significantly poor HRQoL, particularly in mobility, fatigue, and pain. These findings highlight the need for access to targeted therapies, such as burosumab, to improve long-term outcomes and QOL in XLH patients.