DO YOU RECOGNIZE ANY OF THESE SYMPTOMS IN YOUR PATIENTS?

A family living with XLH

- Bone pain, joint pain, and/or joint stiffness
- Fractures and/or pseudofractures
- Muscle pain, weakness, and/or fatigue
- Gait abnormalities
- Short stature
- Osteoarthritis
- Lower limb deformities
- Tooth abscesses and/or excessive dental caries
- Craniosynostosis and Chiari malformation
- Rickets (in children) and osteomalacia
- Sensorineural hearing loss
- Enthesopathy

Most symptoms present in childhood and progress throughout adulthood.[1,2]

Contact an UltraCare representative to discuss how XLH may be impacting your patients.

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COULD IT BE X-LINKED HYPOPHOSPHATEMIA (XLH)?

XLH diagnosis is typically based on clinical and biochemical findings in combination with family history.[3]

Biochemical findings in XLH

Biochemical Test	Test Result[3,4]
Serum phosphorus	↓
$1,25(OH)_2D$	↓ or inappropriately normal
25(OH)D	normal
TmP/GFR	↓
ALP[a]	↑
Serum calcium	normal
Urinary calcium	↓ to normal
PTH	normal to ↑

[a]ALP can be a good marker of skeletal health in children but not necessarily for adults.[3]
$1,25(OH)_2D$, 1,25 dihydroxy vitamin D; 25(OH)D, 25-hydroxy vitamin D (calcifediol); ALP, alkaline phosphatase; PTH, parathyroid hormone; TmP/GFR, ratio of tubular maximum reabsorption of phosphate to glomerular filtration rate.

A diagnosis of XLH can be confirmed with genetic testing for variants in the *PHEX* gene. Contact an UltraCare representative to learn more (see enclosed business card) or visit **XLHLink.com/card**.

REFERENCES: **1**. Carpenter TO, Imel EA, Holm IA, Jan de Beur SM, Insogna KL. A clinician's guide to X-linked hypophosphatemia. *J Bone Miner Res*. 2011;26(7):1381-1388. **2**. Beck-Nielsen SS, Mughal Z, Haffner D, et al. FGF23 and its role in X-linked hypophosphatemia-related morbidity. *Orphanet J Rare Dis*. 2019;14(1):58. **3**. Ruppe MD. X-linked hypophosphatemia. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. **4**. Santos F, Fuente R, Mejia N, Mantecon L, Gil-Peña H, Ordoñez FA. Hypophosphatemia and growth. *Pediatr Nephrol*. 2013;28(4):595-603.

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