HEREDITARY, PROGRESSIVE, AND LIFELONG
X-LINKED HYPOPHOSPHATEMIA (XLH)

INCREASED FGF23 ACTIVITY: A LIFETIME OF IMPACT

TODDLER • ADOLESCENT • YOUNG ADULT • MATURE ADULT
A diagnosis of XLH is typically based on clinical and biochemical findings in combination with family history.

**Family History**

**With Known Family History of XLH**

XLH is inherited in an X-linked dominant pattern. In a family with a history of XLH, screen for other family members. This can help you identify previously undiagnosed individuals.

**Without a Known Family History**

About 20% to 30% of XLH cases are spontaneous. Ask about his/her medical history of short stature, rickets, osteomalacia, and dental abscesses, which may indicate XLH.

**Diagnosis of XLH**

- A diagnosis of XLH can be confirmed through genetic testing for variants of the PHEX gene.

**Clinical Findings**

**Predominant Findings in Children**

Rickets, lower extremity bowing, leg deformities, pain, short stature, and gait disturbances. Confirm skeletal findings through radiography. Other signs and symptoms may also include dental abscesses, craniosynostosis, and Chiari malformations.

**Predominant Findings in Adults**

Adults with XLH may present with osteomalacia manifesting as bone and muscle pain, enthesopathy, fractures, and pseudofractures. Other signs and symptoms may also include waddling gait, dental abscesses, and hearing loss.

**Biochemical Tests**

- Serum phosphorus
- 1,25(OH)2D or inappropriately normal
- 25(OH)D normal
- TmP/GFR normal or slightly increased
- ALP normal
- Serum calcium normal to slightly increased
- Urinary calcium normal to slightly increased
- PTH normal or slightly increased

Other biochemical tests that may be useful for establishing the diagnosis of XLH include serum alkaline phosphatase (ALP) levels and TmP/GFR levels. Alkaline phosphatase can be a good marker of skeletal health in children but not necessarily for adults.

1,25(OH)2D3 = 1,25-dihydroxyvitamin D (calcitriol); 25(OH)D = 25-hydroxyvitamin D (calcifediol); ALP = alkaline phosphatase; PTH = parathyroid hormone; TmP/GFR = ratio of tubular maximum reabsorption of phosphate to glomerular filtration rate; XLH = X-linked hypophosphatemia.
XLH HAS A LIFELONG IMPACT ON PATIENTS

IN PATIENTS WITH XLH, CHRONIC HYPOPHOSPHATEMIA DUE TO INCREASED FGF23 ACTIVITY RESULTS IN POOR SKELETAL, MUSCULAR, AND DENTAL HEALTH AND IMPAIRED PHYSICAL FUNCTION

XLH IS A HEREDITARY, PROGRESSIVE, AND LIFELONG DISEASE

FGF23 IS THE ROOT CAUSE OF XLH

RICKETS AND OSTEOMALACIA ARE THE UNDERLYING SOURCES OF COMPOUNDING AND PROGRESSIVE SYMPTOMS OF XLH

XLH POSES A SIGNIFICANT BURDEN ON THE DAILY LIVES OF CHILDREN AND ADULTS DUE TO IMPAIRED PHYSICAL FUNCTION

FAMILY HISTORY, CLINICAL FINDINGS, AND BIOCHEMICAL TESTS CAN BE USED TO ESTABLISH A DIAGNOSIS OF XLH

Learn more at XLHLink.com

References:
5. Skrinar A, Marshall A, San Martin J, Dvorak-Ewell M. X-linked hypophosphatemia (XLH) impairs skeletal health outcomes and physical function in affected adults. Poster presented at: ENDO 2015; March 5-8, 2015; San Diego, CA.