Investigating XLH: Alina's case†

Case overview

Alina is a 3-year-old female patient who presents with the following:[1,2]

- Genu varum (bowed legs) – most apparent in the femurs
- Short stature (86 cm height, 3rd percentile for her age)
- Waddling gait when walking
- Difficulty running and climbing stairs

Patient history

- Clinical features were apparent at 18 months of age
- No known family history of metabolic bone disease
  - Neither parent exhibits any of Alina's symptoms; both are of average height
- Genetic testing at 26 months of age ruled out the following conditions:
  - Shwachman-Diamond syndrome (SDS)
  - Metaphyseal chondrodysplasia, Schmid type (MCDS)

Laboratory test results

Test (reference range)[3,4‡]	**Results**[4,5]
Serum phosphorus (3.8–6.5 mg/dL)	**2.8 mg/dL**
TmP/GFR (2.9–6.5 mg/dL)	**2.4 mg/dL**
25(OH)D (20–50 ng/mL)	41 ng/mL
ALP (142–335 U/L)	**460 U/L**
Serum calcium (9.3–10.6 mg/dL)	9.6 mg/dL
PTH (15–65 pg/mL)	58 pg/mL

25(OH)D=25-hydroxyvitamin D; ALP=alkaline phosphatase; PTH=parathyroid hormone; TmP/GFR=tubular maximum reabsorption of phosphate corrected for glomerular filtration rate; XLH=X-linked hypophosphatemia.

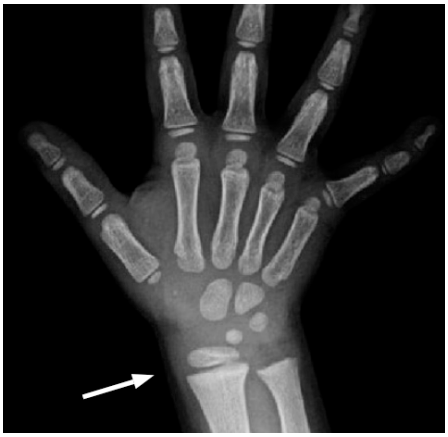
† Fictitious patient. May not be representative of all patients.

‡ Reference ranges may vary based on assay and instrument used. Reference ranges provided by the laboratory conducting the test should be used to ensure accuracy.

XLH LINK

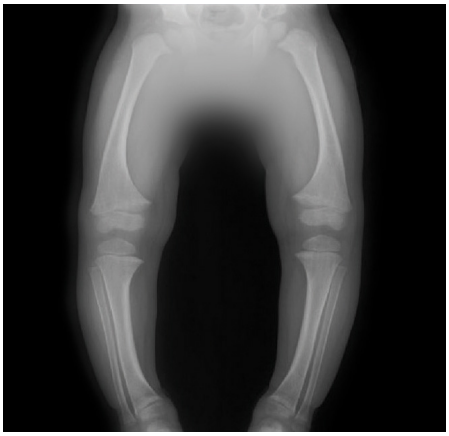
Radiographic evaluation

X-ray 1: Right wrist



Mild metaphyseal fraying at the bilateral distal radius and ulna

X-ray 2: Legs



Femoral and tibial bowing; fraying of the femoral metaphysis

Recommendation from the XLH Guidelines

"In children, a diagnosis of X-linked hypophosphataemia (XLH) should be considered in the presence of clinical and/or radiological signs of rickets, impaired growth velocity and serum levels of phosphate below the age-related reference range associated with renal phosphate wasting and in the absence of vitamin D or calcium deficiency (grade B, moderate recommendation)." — Haffner *et al.*, 2019.[1]

Would you consider genetic testing to confirm a diagnosis of XLH for Alina?

Visit XLHLinkHCP.ca for more information and resources about XLH!

XLH=X-linked hypophosphatemia.

References: 1. Haffner D, *et al. Nat Rev Nephrol.* 2019;15(7):435-455. **2.** Mao M, *et al.* 2020;105(10):3243-3249. **3.** Dahir K, *et al. J Endocr Soc.* 2021;5(9):bvab099. doi:10.1210/jendso/bvab099. **4.** Ruppe MD. X-linked hypophosphatemia. In: Adam MP, Everman DB, Mirzaa GM, *et al.*, eds. GeneReviews®. Seattle (WA): University of Washington, Seattle; February 9, 2012. Updated April 13, 2017. https://www.ncbi.nlm.nih.gov/books/NBK83985/. **5.** Dahir K, *et al. J Endocr Soc.* 2020;4(12):bvaa151.

COMM-CA-RDS-0019 April 2024