Investigating XLH: Devin's case†

Case overview

Devin is a 5-year-old male patient who presents with the following:[1-5]

- Bowed legs
  - X-ray examination confirms bilateral femoral and tibial varus deformities
  - Joint orientation angles further confirm femoral bowing (mLDFA: 93° and 94°; mMPTA: 81° and 80°; in left and right legs, respectively)
- Pain in both of his knees
  - He often takes breaks and sits out during active games
- Impaired growth/short stature (height: 99 cm, 2nd percentile for his age)
- Abnormal gait
- Evidence of rickets in knees and wrists
  - X-rays revealed rachitic changes (see next page)
- Recurrent dental abscesses

Patient history

- Parents noticed slight bowing of the legs and abnormal gait around age 2[6]
- Dental abscesses began appearing after age 3[4]
- His pediatrician initially suspected Blount's disease, but noted that bowing was more prominent in the femurs (rather than the tibiae)
- No known family history of bone disease
  - Neither parent exhibits any of Devin's symptoms; both are of average height

Laboratory test results

Test (reference range)[7,8‡]	**Results**[6,7]
Serum phosphorus (3.7–5.6 mg/dL)	**2.4 mg/dL**
TmP/GFR (2.9–6.5 mg/dL)	**2.2 mg/dL**
25(OH)D (20–50 ng/mL)	36 ng/mL
ALP (142–335 U/L)	**508 U/L**
Serum calcium (9.3–10.6 mg/dL)	9.7 mg/dL
PTH (15–65 pg/mL)	52 pg/mL

25(OH)D=25-hydroxyvitamin D; ALP=alkaline phosphatase; mLDFA=mechanical lateral distal femoral angle; mMPTA=mechanical medial proximal tibial angle; PTH=parathyroid hormone; TmP/GFR=tubular maximum reabsorption of phosphate corrected for glomerular filtration rate; XLH=X-linked hypophosphatemia.

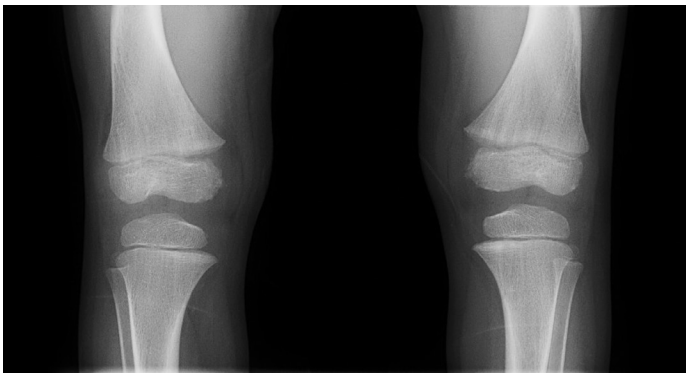
† Fictitious patient. May not be representative of all patients.

‡ 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

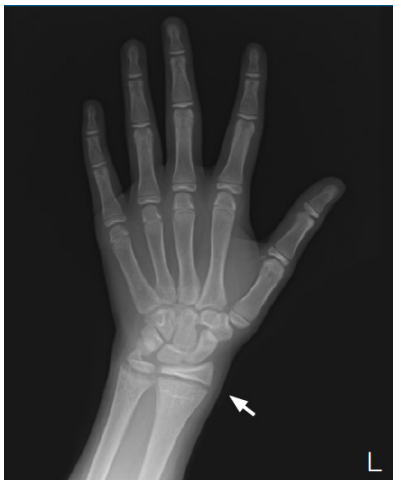
Radiographic evaluation

X-ray 1: Knees



Bilateral fraying of the metaphyses of the distal femurs and proximal tibiae

X-ray 2: Hand



Rachitic changes at the wrist

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.[4]

Would you consider referring Devin to an endocrinologist to help confirm a diagnosis of XLH?

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

XLH=X-linked hypophosphatemia.

References: 1. Luís NM and Varatojo R. *EFORT Open Rev.* 2021;6(6):487-494. **2.** De Cicco A, *et al. Int J Bone Frag.* 2021;1(2):59-66. **3.** Skrinar A, *et al. J Endocr Soc.* 2019;3(7):1321-1334. **4.** Haffner D, *et al. Nat Rev Nephrol*. 2019;15(7):435-455. **5.** Mao M, *et al.* 2020;105(10):3243-3249. **6.** Dahir K, *et al. J Endocr Soc.* 2020;4(12):bvaa151. **7.** 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/. **8.** Dahir K, *et al. J Endocr Soc.* 2021;5(9):bvab099. doi:10.1210/jendso/bvab099.

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