

# Investigating XLH: George's case<sup>†</sup>

## Case overview

George is a 6-year-old patient at the pediatric dentist's office who was referred by a family dentist. He presents with the following:<sup>1-3</sup>

- Unusual abscess at the gums of tooth #14
- No history of trauma
- No history of caries

## Family and patient history<sup>2-6</sup>

- Several members of his family have gum disease
- Mother had numerous root canals and dental issues as a child
- Visibly bowed legs
- Consistent complaints of unspecified "bone pain"
- Short stature (in the 3<sup>rd</sup> percentile for height)

## Clinical exam<sup>2,3,7,8</sup>

Evaluation	Results
Dental caries	No caries observed
Dental abscess	A previously noted abscess on tooth #2 was addressed by extracting the tooth, but a new one on tooth #14 had formed
Enamel health	Hypoplastic enamel observed on several teeth
Overall oral hygiene practices	Good; daily brushing and flossing

XLH=X-linked hypophosphatemia.

<sup>†</sup> Fictitious patient. May not be representative of all patients.

Dental manifestations may present among the first signs of XLH, so dentists can play a role in recognizing symptoms of XLH in children prior to diagnosis<sup>4</sup>

## Photographic and radiographic evaluation

Clinical photo



Arrow points to inflamed gums due to endodontic abscess.

Periapical radiograph



Radiographic image of same tooth.

Images provided courtesy of Juan F. Yepes.

## Recommendation from the XLH Guidelines<sup>2</sup>

“Patients should have at least twice-yearly dental examinations after tooth eruption, orthodontic evaluation around the age of 12 years and an extended dental evaluation with transition to adult care. The number of dental abscesses and episodes of acute oral infections (including maxillofacial cellulitis) should be recorded at each visit (as these are indirect indices of impaired tooth mineralization).”



Would you consider referring George to a pediatric endocrinologist to help confirm a diagnosis of XLH?



Visit [XLHLinkHCP.ca](https://www.xlhlinkhcp.ca) for more information and resources about XLH!

XLH=X-linked hypophosphatemia.

**References:** 1. Duplan MB, et al. *Int J Bone Frag.* 2021;1(2):74-79. 2. Haffner D, et al. *Nat Rev Nephrol.* 2019;15(7):435-455. 3. Xinyang J, et al. *BDJ Open.* 2023;9(2). 4. Okawa R, Nokano K. *Endocrines.* 2022;3(4):654-664. 5. Glorieux FH, et al. *Orphanet J Rare Dis.* 2022;17(1):30. 6. Mao M, et al. *Clin Endocrinol Metab.* 2020;105(10):3243-3249. 7. Zambrano M, et al. *Oral Surgery, Oral Medicine, Oral Pathology, Oral Radiology, and Endontology.* 2003;95(6):705-709. 8. Lee B-N, et al. *RDE.* 2017;42(2):146-151.