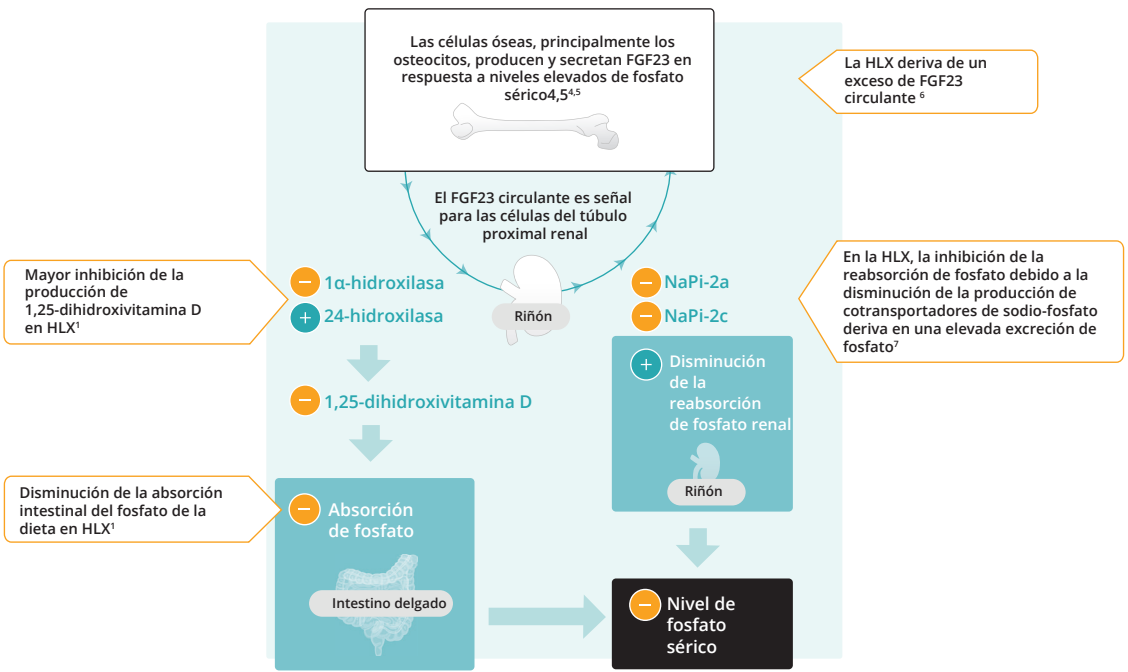
La hipofosfatemia ligada al cromosoma X (HLX) es un trastorno esquelético crónico y progresivo[1,2]

La HLX está caracterizada por pérdida renal de fosfato, que está causada por un exceso en la producción del factor de crecimiento fibroblástico 23 (FGF23)[1,2]

En personas normales, el FGF23 ayuda a mantener la homeostasis del fosfato, que es fundamental para una salud esquelética permanente[3]

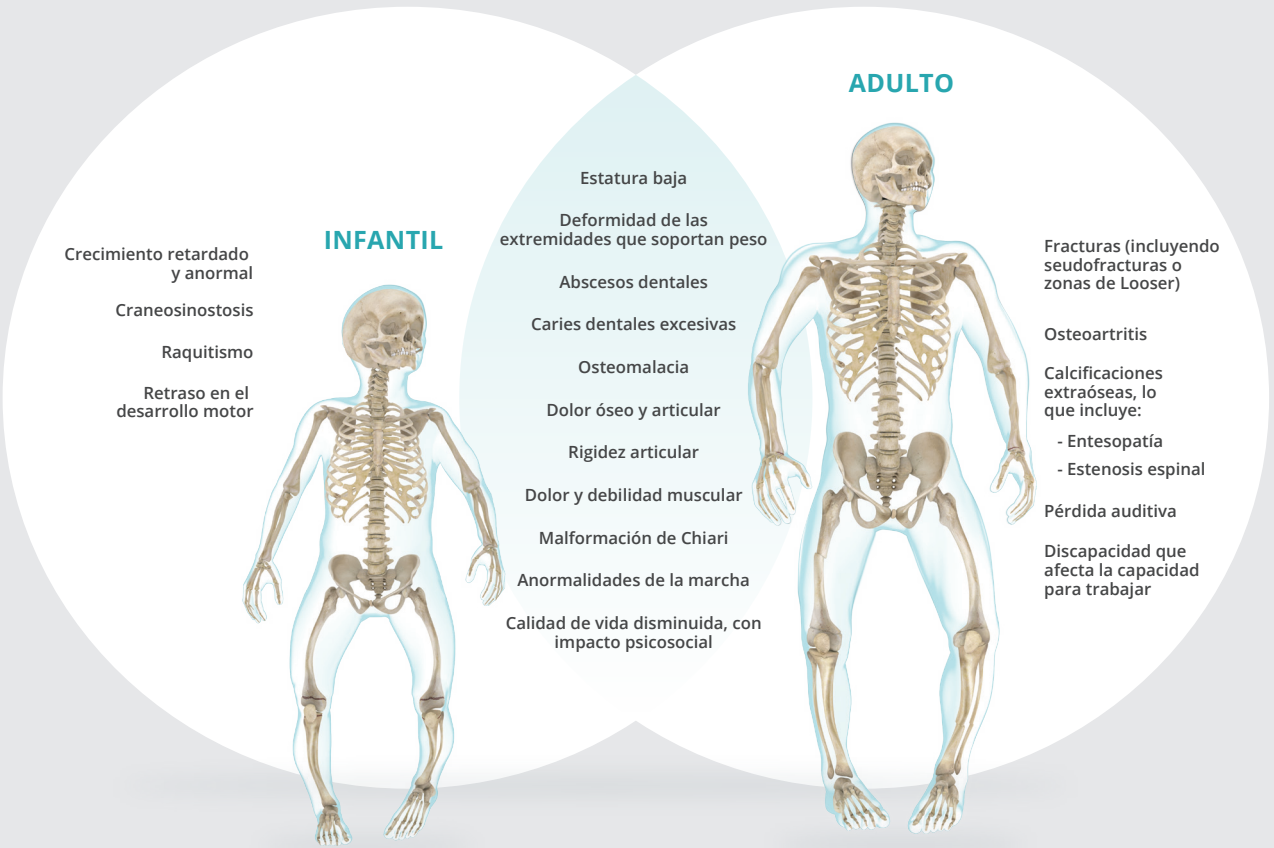


En pacientes con HLX, el exceso de FGF23 deriva en hipofosfatemia crónica causada por lo siguiente[2,3,8]:

- Pérdida renal de fosfato
- Disminución de la absorción intestinal del fosfato

Esto deriva en una mineralización deficiente de los huesos y los dientes[9]

Las consecuencias de la HLX tienen un impacto constante en la salud esquelética[6,10-17]



Las manifestaciones clínicas en los adultos con HLX surgen como consecuencia de complicaciones no resueltas de la HLX durante la infancia y/o una enfermedad activa persistente[11,13]

REFERENCIAS: 1. Martin A, Quarles LD. Evidence for FGF23 involvement in a bone-kidney axis regulating bone mineralization and systemic phosphate and vitamin D homeostasis. *Adv Exp Med Biol.* 2012;728:65-83. **2.** 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. **3**. Penido MG, Alon US. Phosphate homeostasis and its role in bone health. *Pediatr Nephrol.* 2012;27(11):2039-2048. **4.** Riminucci M, Collins MT, Fedarko NS, et al. FGF-23 in fibrous dysplasia of bone and its relationship to renal phosphate wasting. *J Clin Invest.* 2003;112(5):683-692. **5.** Ferrari SL, Bonjour J-P, Rizzoli R. Fibroblast growth factor-23 relationship to dietary phosphate and renal phosphate handling in healthy young men. *J Clin Endocrinol Metab.* 2005;90(3):1519-1524. **6.** Che H, Roux C, Etcheto A, et al. Impaired quality of life in adults with X-linked hypophosphatemia and skeletal symptoms. *Eur J Endocrinol.* 2016;174(3):325-333. **7.** Gattineni J, Bates C, Twombley K, et al. FGF23 decreases renal NaPi-2a and NaPi-2c expression and induces hypophosphatemia in vivo predominantly via FGF receptor 1. *Am J Physiol Renal Physiol.* 2009;297(2):F282-F291. **8.** Ruppe MD. X-linked hypophosphatemia. In: Pagon RA, Adam MP, Ardinger HH, et al, eds. *Gene Reviews.* https://www.ncbi.nlm.nih.gov/books/NBK83985/. Accessed October 20, 2017. **9.** Pettifor JM. What's new in hypophosphataemic rickets? *Eur J Pediatr.* 2008;167(5):493-499. **10.** Carpenter TO. Primary disorders of phosphate metabolism. In: De Groot LJ, Chrousos G, Dungan K, et al, eds. *Endotext [internet].* South Dartmouth, MA: MDText.com. 2014;1-56. **11.** Linglart A, Biosse-Duplan M, Briot K, et al. Therapeutic management of hypophosphatemic rickets from infancy to adulthood. *Endocr Connect.* 2014;3(1):R13-R30. **12.** Linglart A, Dvorak-Ewell M, Marshall A, et al. Impaired mobility and pain significantly impact the quality of life of children with X-linked hypophosphatemia (XLH). Poster presented at: ICCBH 2015 Salzburg, Austria. **13.** 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: Endocrine Society's 97th Annual Meeting and Expo, March 5-8, 2015. San Diego, CA. **14.** Veilleux LN, Cheung M, Ben Amor M, Rauch F. Abnormalities in muscle density and muscle function in hypophosphatemic rickets. *J Clin Endocrinol Metab.* 2012;97(8):E1492-E1498. **15.** Data on file, Ultragenyx, Inc. **16.** Looser zones. Radiopaedia Web site. https://radiopaedia.org/articles/looser-zones-1. Accessed October 9, 2017. **17.** Zivičnjak M, Schnabel D, Billing H, et al. Age-related stature and linear body segments in children with X-linked hypophosphatemic rickets. *Pediatr Nephrol.* 2011;26(2):223-231.

ultragenyx pharmaceutical

 MRCP-KRN23-00295 04/18

KYOWA KIRIN