

Connect the symptoms.

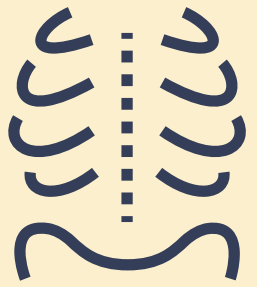
Should you test for X-linked hypophosphatemia (XLH)?

Learn more about the signs and symptoms of XLH and how to accurately diagnose it

What is XLH?

X-linked hypophosphatemia (XLH) is a rare, lifelong, progressive disease characterized by hypophosphatemia. A variant in the *PHEX* gene causes excess fibroblast growth factor 23 (FGF23) activity, which results in phosphorus wasting leading to chronic hypophosphatemia.^{1,2}

XLH is a progressive disease that can cause¹⁻⁴:

		
Skeletal defects	Muscle dysfunction	Dental issues



XLH impacts children and adults throughout their lives and may require lifelong management.¹⁻⁴



ISLA, LIVING WITH XLH

Symptoms of XLH are nonspecific, which may lead to misdiagnosis

Symptoms such as...

- Poor skeletal, muscular, and dental health²⁻⁴
- Impaired physical function⁵

...often result in a misdiagnosis. Examples include^{1,6}:

- Other hereditary rickets
- Nutritional rickets
- Osteomalacia
- Hypophosphatasia
- Physiologic bowing

Misdiagnosis of XLH may lead to inappropriate disease management, disease progression, and potential increased symptom severity.^{1,6-8}

In order to establish an accurate diagnosis of XLH, the results of genetic and FGF23 testing can be considered, in addition to^{1,2,7}:



Clinical/radiographic findings



Family history



Biochemical tests

The value of diagnosis

With an early and accurate diagnosis of XLH, patients can:

Learn more

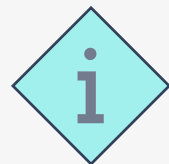
Educate patients to help them understand their condition and appropriate management options

Be connected to resources

Provide patients with the necessary support, including genetic counseling for appropriate patients, to help them navigate their condition

Reduce the risk of further damage

With knowledge and individualized care, patients can potentially limit the progression of XLH over time



Kyowa Kirin is offering no-charge genetic and FGF23 testing for patients who are being evaluated for a possible diagnosis of XLH.*

*See Terms of Use.

ARLENE AND HER DAUGHTER GINA,
BOTH LIVING WITH XLH



XLH

LINK

Diagnose XLH accurately with genetic and FGF23 testing

Confirm XLH:

Using gene panels that include *PHEX* may accelerate the confirmatory diagnosis of phosphorus wasting disorders like XLH

Learn more about [sponsored genetic testing](#)

Check for intact FGF23 levels, which are often elevated in patients with XLH

Request a sponsored [FGF23 test kit*](#)



XLH is caused by X-linked dominant variants in the *PHEX* gene^{1,6}



Symptoms of XLH are nonspecific and often result in misdiagnosis, which can lead to disease progression and increased symptom severity^{1,6-8}



Early and accurate diagnosis and management can potentially limit the worsening of patient symptoms over time^{1,6,7}



Genetic counseling can help patients and families navigate life with a hereditary condition like XLH

*See Terms of Use.

XLH

LINK



For more information,
visit XLHLinkHCP.com

Terms of Use

Kyowa Kirin has partnered with Invitae Corp. and collaborated with Mayo Clinic Laboratories to offer sponsored, no-charge testing to patients who are being evaluated for a possible diagnosis of XLH or TIO. The Kyowa Kirin Sponsored Hypophosphatemia Program is intended to improve patient safety and quality of care by shortening the time to an accurate diagnosis, facilitate prompt confirmatory testing, and help patients with XLH and TIO meet payor coverage requirements. Use of or participation in the Kyowa Kirin Sponsored Hypophosphatemia Program does not create any obligation to use, prescribe, or recommend any Kyowa Kirin products or services. The Program is only available to patients that meet certain eligibility requirements and for whom the patient's health care provider has determined that testing is clinically appropriate. No patient or health care provider may seek reimbursement for testing or counseling services provided under the Kyowa Kirin Sponsored Hypophosphatemia Program from any third party, including but not limited to, any government health care programs.

References:

1. 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. doi:10.1002/jbmr.340 **2.** 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/> **3.** Linglart A, Bissio-Duplan M, Briot K, et al. Therapeutic management of hypophosphatemic rickets from infancy to adulthood. *Endocr Connect.* 2014;3(1):R13-R30. doi:10.1530/EC-13-0103 **4.** Dahir K, Roberts MS, Krolczyk S, Simmons JH. X-linked hypophosphatemia: a new era in management. *J Endocr Soc.* 2020;4(12):bvaa151. doi:10.1210/jendso/bvaa151 **5.** Orlando G, Bubbear J, Clarke S, et al. Physical function and physical activity in adults with X-linked hypophosphatemia. *Osteoporos Int.* 2022;33(7):1485-1491. doi:10.1007/s00198-022-06318-w **6.** Rush ET, Johnson B, Aradhya S, et al. Molecular diagnoses of X-linked and other genetic hypophosphatemias: results from a sponsored genetic testing program. *J Bone Miner Res.* 2022;37(2):202-214. doi:10.1002/jbmr.4454 **7.** Trombetti A, Al-Daghri N, Brandi ML, et al. Interdisciplinary management of FGF23-related phosphate wasting syndromes: a Consensus Statement on the evaluation, diagnosis and care of patients with X-linked hypophosphatemia. *Nat Rev Endocrinol.* 2022;18(6):366-384. doi:10.1038/s41574-022-00662-x **8.** Hamilton AA, Faitos S, Jones G, Kinsley A, Gupta RN, Lewiecki EM. Whole body, whole life, whole family: patients' perspectives on X-linked hypophosphatemia. *J Endocr Soc.* 2022;6(8):bvac086. doi:10.1210/jendso/bvac086