Nonsponsored US Commercial Diagnostic Laboratories Offering *PHEX* Gene Next-Generation Sequencing (NGS) *AND* Deletion/Duplication Detection*

Molecular testing of the *PHEX* gene may help to confirm a diagnosis of X-linked hypophosphatemia (XLH).[1]

Suggested CPT Code: 81406x1 (Sequencing)

Suggested CPT Code: 81479x1 (Deletion/Duplication)

Use of the above CPT Codes does not guarantee reimbursement from a healthcare plan or provider.

Invitae

Phone: (800) 436-3037
Fax: (415) 276-4164
Email: www.invitae.com/en/contact/
Website: www.invitae.com

Test Code: 72038
Test Name: Invitae X-Linked Hypophosphatemia Test
Estimated Turnaround Time: 10-21 calendar days (14 days on average)

GeneDx

Phone: (888) 729-1206
Fax: (201) 421-2010
Email: zebras@genedx.com
Website: www.genedx.com

Test Code: 1861E
Test Name: Hypophosphatemic Rickets, X-linked, *PHEX* Gene Sequencing and Del/Dup
Estimated Turnaround Time: 3 weeks

Blueprint Genetics

Phone: (650) 452-9340 Ext. 0
Fax: (650) 446-7790
Email: support.us@blueprintgenetics.com
Website: blueprintgenetics.com

Test Code: S01406
Test Name: *PHEX* Single Gene Test Plus Analysis (Sequence and Del/Dup [CNV])
Estimated Turnaround Time: 3-4 weeks

Cost to patient may vary with insurance coverage. In case of hardship, 501(c)(3) Foundations Donations may be able to provide financial assistance that covers incidental medical expenses (IMEs) related to the patient's disease. The Assistance Fund can help determine your patient's eligibility for financial assistance. For further information, please call (855) 341-1510.

This list is offered only as an optional resource to locate providers offering both *PHEX* gene NGS and *PHEX* gene deletion/duplication detection. This list may not be exhaustive and may not provide the most up-to-date information. Inclusion on this list does not imply an endorsement by or association with Ultragenyx Pharmaceutical Inc. A more comprehensive list of labs offering *PHEX* testing can be found in the Labs tab on the GTR:Genetic Testing Registry site at https://www.ncbi.nlm.nih.gov/gtr/.

Sequence analysis *AND* deletion/duplication analysis maximizes the chance of making an accurate molecular genetic diagnosis. Sequence analysis *OR* deletion/duplication analysis may be a more cost-effective option if a family member already has an established molecular genetic diagnosis of XLH.

Additional providers may offer only Sanger sequencing of *PHEX* (not NGS), or may offer gene panel assays that include *PHEX*, but are often more expensive and may not include deletion/duplication testing.

Information current as of March 2020.

Reference:

1. 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 19, 2018.

*Please use this list if your institution does not allow sponsored testing. If your institution does allow sponsored testing, note that Ultragenyx has partnered with Invitae to offer sponsored genetic testing. For more information, visit www.invitae.com/en/hypophosphatemia/.

Nonsponsored US Commercial Diagnostic Laboratories Offering an FGF23 ELISA Assay*

Assessment of plasma FGF23 levels may help to confirm a diagnosis of X-linked hypophosphatemia (XLH), as characterized, in part, by low serum phosphate levels.[1,2]

Suggested CPT Code: 83520

Use of the above CPT Code does not guarantee reimbursement from a healthcare plan or provider.

Mayo Clinic Laboratories

Phone: (800) 533-1710
Fax: (507) 284-1759
Email: mcl@mayo.edu
Website: www.mayocliniclabs.com

Test Code: IFG23
Test Name: Intact Fibroblast Growth Factor 23, Serum
Assay Type: Intact FGF23
Estimated Turnaround Time: 7 days

Nonsponsored US Commercial Diagnostic Laboratories Offering a Tubular Reabsorption of Phosphorus Test

Assessment of the ratio of the maximum rate of tubular phosphate reabsorption to the glomular filtration rate (TmP/GFR) may also help to confirm a diagnosis of XLH.[1]

Suggested CPT Codes: 82565, 84100, 84105

Use of the above CPT Codes does not guarantee reimbursement from a healthcare plan or provider.

Mayo Clinic Laboratories

Phone: (800) 533-1710
Fax: (507) 284-1759
Email: mcl@mayo.edu
Website: www.mayocliniclabs.com

Test Code: RTRP
Test Name: Tubular Reabsorption of Phosphorus, Random Urine and Serum
Estimated Turnaround Time: Same day/1 day

Cost to patient may vary with insurance coverage. In case of hardship, 501(c)(3) Foundations Donations may be able to provide financial assistance that covers incidental medical expenses (IMEs) related to the patient's disease. The Assistance Fund can help determine your patient's eligibility for financial assistance. For further information, please call (855) 341-1510.

This list is offered only as an optional resource to locate providers offering an FGF23 ELISA or TmP/GFR assay. This list may not be exhaustive and may not provide the most up-to-date information. Inclusion on this list does not imply an endorsement by or association with Ultragenyx Pharmaceutical Inc.

Information current as of March 2020.

References:

1. 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 19, 2018.
2. Jonsson KB, Zahradnik R, Larsson T, et al. Fibroblast growth factor 23 in oncogenic osteomalacia and X-linked hypophosphatemia. *N Engl J Med.* 2003;348(17):1656-1663.

*Please use this list if your institution does not allow sponsored testing. If your institution does allow sponsored testing, note that Ultragenyx has partnered with the Mayo Clinics to offer sponsored FGF23 testing. For more information, contact UltraCare Support at (888) 756-8657 or online at ultracaresupport.com.

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