PHEX MOLECULAR TESTING

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 Codes: 81406x1 (Sequencing)
81479x1 (Deletion/Duplication)

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

Connective Tissue Gene Tests (CTGT)

Phone: (484) 244-2900
Fax: (484) 244-2904
Email: inquiries@ctgt.net
Website: ctgt.net

Test Code: 1636
Test Name:
Rickets, Hypophosphatemic, X-linked Dominant, NGS/Del Dup Comprehensive
Estimated Turnaround Time: 2-4 weeks

GeneDx

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

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 October 2018.

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.

FGF23 ELISA ASSAY

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 Medical Laboratories

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

Test Code: FGF23
Test Name:
Fibroblast Growth Factor 23 (FGF23), Plasma
Assay Type: C-terminal FGF23
Estimated Turnaround Time: 8 days

Quest Diagnostics

Phone: 866-MYQUEST (866-697-8378)
Website: www.questdiagnostics.com

Test Code: 91931
Test Name:
FGF 23 (Fibroblast Growth Factor 23)
Assay Type: C-terminal FGF23
Estimated Turnaround Time: 7 days

LabCorp

Website: www.labcorp.com

Test Code: 004380
Test Name:
Fibroblast Growth Factor 23
Assay Type: C-terminal FGF23
Estimated Turnaround Time: Not provided

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 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.

We are not aware of any commercial laboratories currently providing an intact FGF23 assay (eg, Kainos assay).

Information current as of October 2018.

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.

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