XLH Confirmatory Testing

SPONSORED NO-CHARGE GENETIC TESTING TO CONFIRM XLH DIAGNOSIS FOR TREATMENT COVERAGE

What is XLH Confirmatory Testing?

Some insurers require verification of XLH diagnosis before determining patient eligibility for treatment coverage. XLH Confirmatory Testing is a sponsored program that provides genetic confirmation of XLH diagnosis.

Who is eligible for XLH Confirmatory Testing?

Patients are eligible if they are aged **6 months or older** and have a **completed UltraCare® Start Form for XLH**. Testing is available to all eligible patients at no charge.

How do I submit a patient test?

STEP 1
Complete and print both pages of the **Invitae test requisition form**.

STEP 2
Obtain a blood, saliva, or buccal sample from your patient using the provided Invitae kit.

STEP 3
Mail the form and patient sample using the provided packaging and prepaid label.

Questions about XLH Confirmatory Testing?

Contact UltraCare at **1-888-756-8657** or online at **ultracaresupport.com**.

It is strongly encouraged to provide medical history, including clinical and biochemical findings, and family history information with the form and patient sample, when available. This information is useful for variant interpretation. Test results are usually available within 10 to 21 calendar days. You will be notified via email or fax to access results through Invitae's secure site. Obtain patient permission before sharing test results with the patient's insurance company.

Complete the following fillable test requisition form, print it out, and enclose it with the patient sample. The form may also be submitted via fax to **415-276-4164** or online at **invitae.com/hypophosphatemia**.

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Sponsored by: ultragenyx pharmaceutical

INVITAE **HYPOPHOSPHATEMIA**

ORDER ID
For Invitae internal use only

Requisition Form
Hypophosphatemia Sponsored Testing Program
TRF942-5

This requisition form can be used to submit an order for the **Hypophosphatemia Program**, a complimentary testing program for **genetic hypophosphatemic disorders** brought to you by **Ultragenyx Pharmaceutical, Inc.**

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

ORDERING OPTIONS

1. HYPOPHOSPHATEMIA PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.

REQUIRED: You must select below the appropriate eligibility criteria for this patient.

The patient must be aged 6 months or older and meet one of the following criteria below (select one or more):

- ◯ Has completed the UltraCare® Start Form for XLH
- ◯ Has a previous diagnosis related to hypophosphatemia*
- ◯ Has a family member with a confirmed XLH diagnosis
- ◯ Exhibits **TWO or more** of the following clinical signs and/or symptoms (select two or more):
  - ◯ Muscle pain, weakness, and/or fatigue
  - ◯ Lower limb deformities
  - ◯ Fractures/pseudo-fractures
  - ◯ Tooth abscesses and/or excessive dental caries
  - ◯ Bone, joint pain, and/or joint stiffness
  - ◯ Short stature
  - ◯ Gait abnormalities

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional charge. Relatives do not need to meet the eligibility criteria listed above. Learn more at www.invitae.com/family.

*Hypophosphatemic disorders are also referred to as X-linked hypophosphatemia, X-linked hypophosphatemic rickets, hereditary hypophosphatemic rickets, familial hypophosphatemic rickets, vitamin D-resistant rickets (VDRR), vitamin D-resistant osteomalacia, X-linked vitamin D-resistant rickets, hypophosphatemic rickets, hypophosphatemic vitamin D-resistant rickets (HPDR), genetic rickets, or familial hypophosphatemia.

PATIENT INFORMATION

First name | MI | Last name

Date of birth (MM/DD/YYYY) | Biological sex ◯ M ◯ F | MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other: _____

Phone | Email address (report access after clinician releases)

Address | City

State/Prov | ZIP/Postal code | Country

Ship a kit to this patient (optional) by faxing this form to Client Services at 415-276-4164

Kit type: ◯ Buccal swab kit ◯ Saliva kit

Ship to: ◯ Address above ◯ Alternate address: _____

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) -OR- Buccal Swab -OR- Saliva (Oragene™) -OR- Assisted Saliva

We are unable to accept blood/buccal/saliva from patients with:
- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Specimen collection date (MM/DD/YYYY): ☐☐ ☐☐ ☐☐☐☐
If not provided, the day before specimen receipt will be used

Special cases: ◯ History of/current hematologic malignancy in patient

CLINICIAN INFORMATION

Organization name

Phone | Fax

Address | City

State/Prov | ZIP/Postal code | Country

Primary clinical contact name (if different from ordering provider) | NPI

Primary clinical contact email address (for report access)

Ordering provider (select one ordering provider by marking the checkbox before the name)

	Name	NPI	Email address (for report access)
◯			
◯			
◯			
◯			

Additional clinical or laboratory contacts (optional, to share access to order online)

◯ **Share this order with the primary clinical contact's default clinical team, manage at invitae.com**

Name | Email address (for report access)

Name | Email address (for report access)

POST-TEST GENETIC COUNSELING (please review)

Ultragenyx sponsors post-test genetic counseling, regardless of test result type, at no additional charge to the patient through InformedDNA, an independent genetic counseling service. Please check the box below if you would like InformedDNA to contact your patient for genetic counseling once the patient's test is complete.

◯ **I authorize Invitae to grant InformedDNA (patientcare@InformedDNA.com) access to the patient's Invitae record for the purposes of providing genetic counseling services**

INVITAE PARTNER CODE XLH

INVITAE **HYPOPHOSPHATEMIA**

Hypophosphatemia Sponsored Testing Program TRF942-5

CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis	Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?† ◯ Yes ◯ No

Provide details in the required clinical history questions (if applicable).

† Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

CLINICAL HISTORY

Biochemical markers (select all that apply): It is strongly encouraged to include a copy or the values of abnormal results, when available, for this individual or affected family members. This information is useful for variant interpretation.

	Patient value/reference range
◯ Reduced serum phosphate (<LLN)	_____ /_____
◯ Reduced TmP/GFR (<LLN)	_____ /_____

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

*To have the presence or absence of specific variants commented on in this patient's report, provide the details below. For gene-specific family follow-up see **Note** under Test Selection.*

Was the proband (individual with variant) tested at Invitae? ◯ Yes, Invitae Order ID: RQ# _____ ◯ No: Attach copy of lab results (*required*)

Variant(s) *(e.g. GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.*		**This patient's relationship to proband:**
		◯ Parent ◯ Sibling ◯ Grandchild ◯ Child ◯ Self ◯ Other: _____

TEST SELECTION – Select option 1 or 2 below:

◯ 1. HYPOPHOSPHATEMIA PROGRAM – Indicate test(s) to be performed below:

Test code	Test name	# of genes	Gene list
◯ 72039	Invitae Hypophosphatemia Panel	17	ALPL, CLCN5, CTNS, CYP2R1, CYP27B1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR

◯ 2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING *For relatives of a program participant ('proband') who received a Pathogenic/Likely Pathogenic result or approved VUS.*

Proband's Invitae Order ID: RQ# _____	**This patient's relationship to proband:** ◯ Parent ◯ Sibling ◯ Grandchild ◯ Child ◯ Other: _____	**Gene(s) to be tested in this patient:**

***NOTE**: The presence or absence of all variants identified in the proband for the gene(s) ordered for gene-specific family follow-up will be commented on in this patient's report unless a limited selection is specified in the **Requested Variants** section above. Invitae will report any Pathogenic/Likely Pathogenic variants found in this patient for the gene(s) ordered.*

Invitae continually updates its panels based on the most recent evidence. If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. Test IDs containing add-on codes will include the original panel as well as the add-on.

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/forms). In connection with the Program the Patient has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated) and has been informed that deidentified Patient data may be used and shared with third parties, for research and commercial purposes and, in the U.S., to contact their medical professional. For orders originating in Canada, the Patient has been informed that their personal information and specimen will be transferred to and processed in the U.S. and that de-identified Patient data may be used and shared for research and commercial purposes in the U.S. The medical professional warrants that he/she will not seek reimbursement for this no-charge test from any third party, including but not limited to federal healthcare programs. The medical professional also hereby acknowledges that organization and clinician contact information provided in the order may be shared with third parties, including commercial organizations, that may contact the medical professional directly in connection with the Program, and that they have made the Patient aware that de-identified Patient data may be used and shared with such third parties, for purposes which include contacting their medical professional directly in connection with the Program. A list of third party partners may be provided upon request. I attest that I am authorized under applicable state law to order this test.

Medical professional signature (required)	**Date (MM/DD/YYYY)**