X-linked Hypophosphatemia (XLH):

SPONSORED GENETIC COUNSELING FOR PATIENTS

How to make an appointment with a genetic counselor

If you or your child have been diagnosed with XLH or you have a family member with XLH, consider connecting with a genetic counselor. For additional information about this genetic counseling program, visit **InformedDNA.com/Ultragenyx**. **This sponsored service is offered at no charge to you.**

1 Schedule an appointment

Individuals with XLH and their family members can call InformedDNA directly at 1-888-744-8952 to request an appointment. You do not need a referral from your doctor. Some doctors may send a referral; in that case, InformedDNA will contact you via email and phone to schedule an appointment. The email will include an option to schedule the appointment online. Day, evening, and weekend appointments are available. Interpreter services are available for non-English speakers.

2 Provide your consent and family history online

InformedDNA has a secure online Patient Portal. You will receive portal login details via email. The portal is where you can schedule your appointment, provide your consent to participate in genetic counseling (consent is required before you can speak with a genetic counselor), and complete your family history. If you do not have online access, **InformedDNA** will make alternative arrangements for you.

3 Meet with your genetic counselor

At the scheduled time, your genetic counselor will call you to go over your test results, if available, and what they mean for your extended family and discuss any questions you may have. Feel free to have a family member or friend join you on the call. After the call your counselor will send you and your doctor a summary report. If you need any future support, you'll be connected to the same counselor. **Genetic counseling is available in English, with interpreter services for non-English speakers.**

What happens during my phone appointment with the genetic counselor?

YOUR GENETIC COUNSELOR WILL:

- **Review the clinical features and inheritance pattern of XLH and your genetic test results, if available**
- **Discuss options to manage your health and connect family members with XLH experts for medical management**
- **Review X-linked inheritance and identify which family members may be at risk to have XLH**
- **Provide guidance for how to speak with family members who may be at risk of having XLH**

InformedDNA is the authority on the appropriate use of genetic testing. It leverages the expertise of the largest full-time staff of independent, board-certified genetics specialists in the US to help ensure that health plans, hospitals, employers, pharmaceutical companies, clinicians, and patients all have access to the highest quality genetic services. Key offerings include clinical genetic counseling, genetic testing utilization management, genetic testing payment integrity, and expert support for genetics-based clinical trials. For more information: www.InformedDNA.com

Counseling by: InformedDNA® Genetics, Decoded.

Sponsored by: ultragenyx pharmaceutical

NCQA CERTIFIED UTILIZATION MANAGEMENT

Diagnosed with XLH?

WE'RE HERE TO HELP

What is X-linked hypophosphatemia (XLH)?

XLH is a rare genetic disorder that affects the bone, muscle, and dental health of approximately 3,000 children and 9,000 adults in the United States. "X-linked" means the disorder can be passed from biological parent to child on the X chromosome and "hypophosphatemia" means having unusually low levels of the mineral phosphate in the blood. XLH is known by many different names.

Other names for XLH:

- X-linked hypophosphatemic rickets
- Familial hypophosphatemic rickets
- Hereditary 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)
- X-linked rickets
- Genetic rickets
- Familial hypophosphatemia

What causes XLH?

XLH is caused by mutations in a gene called *PHEX*. *PHEX* mutations lead to hypophosphatemia. Because bones need adequate amounts of phosphate to grow properly and become strong, patients with XLH may have poor bone health for their entire lives. XLH can also cause abnormalities in skeletal muscles and teeth.

What are the signs and symptoms of XLH?

People with XLH can experience a variety of symptoms, including slow or delayed growth (short stature), bone pain, muscle pain, muscle weakness, leg bowing, fractures, osteoarthritis, skeletal or soft tissue calcification, and tooth abscesses. Because symptoms can vary among people with the same condition, patients may be misdiagnosed or go undiagnosed for some time before discovering they have XLH.

Why should I choose genetic counseling?

Getting diagnosed with a genetic disorder can be overwhelming. Genetic counselors can help you manage your or your child's condition, identify family members at risk, and assist with family planning.

How much does genetic counseling cost?

This sponsored service is offered free of charge to eligible patients aged 6 months and older who have been diagnosed with XLH and their family members.

FAQs ABOUT XLH

Q: Are my children also at risk for XLH?

A: The genetic mutation that causes XLH can be passed from biological parent to child: children of mothers with XLH have a 50% chance of inheriting the mutation, while fathers with XLH will pass the mutation on to all of their daughters and none of their sons. Knowing that you have XLH can help explain symptoms you or your children may have. Your genetic counselor can provide a more detailed explanation of why XLH is inherited differently from mothers and fathers.

Q: My doctor has diagnosed me with XLH but no one else in my family has the same symptoms that I am experiencing. Is that possible?

A: Yes. There are many signs and symptoms of XLH, so it is possible for family members who also have XLH to experience different symptoms.

It is also possible that you are one of the 20%-30% of people with XLH who did not inherit the genetic mutation responsible for XLH; rather, there was a spontaneous mutation that led to your XLH. Because XLH is rare, it is unlikely that your blood relatives would have XLH if your case was caused by a spontaneous mutation.

Questions about being diagnosed with XLH? Are you or your child eligible for this sponsored genetic counseling program? **Learn more** at **InformedDNA.com/Ultragenyx** or **call InformedDNA** at **1-888-744-8952**.

InformedDNA®
Genetics, Decoded.

Genetic Counseling Referral

Date ___ ___ _____

Patient Information* *(*all fields are required. Mark "No Email" if the patient doesn't have email.)*

Name:_____ Date of Birth:___ ___ _____

1

PREFERRED ☐Phone:_____ SECONDARY ☐Phone:_____ Email:_____

Language interpreter needed: ☐Spanish ☐Other:_____

Billing

2 Bill to Ultragenyx Pharmaceutical, Inc.

Reason for Referral

This referral form can be used to submit a request for sponsored genetic counseling and genetic testing, a complimentary program brought to you by Ultragenyx Pharmaceutical, Inc. Patients must meet the eligibility requirements for the program. To submit referrals for genetic counseling outside of this program, please order through InformedDNA's website: www.InformedDNA.com

Program Eligibility.

3

Patient **must be age 6 months or older** and:

☐ diagnosed with X-linked hypophosphatemia (XLH)

OR

☐ have a family member diagnosed with X-linked hypophosphatemia (XLH)

NOTE: All information provided in this Genetic Counseling Referral form will remain confidential and shall only be used for the purpose of providing referrals related to genetic counseling. Genetic counseling appointments are sponsored by Ultragenyx Pharmaceutical, Inc. All genetic counseling will be provided by an independent genetic counselor assigned solely by InformedDNA. Ultragenyx does not have any role in the selecting of the genetic counselor.

Physician Information

4

_____
Medical Center/Practice

_____
Referring Provider

|_|_|_|_|_|_|_|_|_|_|
NPI

_____
Practice Contact

_____ _____
Phone Fax (required)

_____
E-mail

_____
Address

_____ _____ _____
City State Zip

_____
Referring Provider's Signature

By submitting this referral form I, the referring provider listed above, am (1) requesting my patient receive genetic counseling by an InformedDNA genetic counselor; and if appropriate (2) authorizing InformedDNA's genetic counselors to facilitate genetic testing using the Hypophosphatemia Panel through Invitae lab, on my behalf utilizing my name and NPI. Test results will be reported to both the genetic counselor at InformedDNA and to me. The genetic counselor will disclose the test results to the patient and family.

Fax completed form to:

5 **760-308-6324**

Questions about our services?

888-744-8952

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