

X-linked Hypophosphatemia (XLH):

SPONSORED GENETIC COUNSELING FOR PATIENTS



How to make an appointment with your genetic counselor

If your doctor has received genetic or biochemical test results confirming that you or your child have XLH, consider connecting with a genetic counselor.

This sponsored service is offered at no cost to you.



1 Contact your doctor to submit a Genetic Counseling Referral Form

In order to set up an appointment, your doctor will first need to complete a Genetic Counseling Referral Form and provide your XLH test results to InformedDNA. Either you or your doctor can **call InformedDNA at 1-888-744-8952** to get this started.



2 Schedule an appointment

Once the referral from your doctor is received, 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.



3 Provide your consent and family history online

Once InformedDNA receives the referral form from your doctor, you will receive an email with information on how to access InformedDNA's secure online Patient Portal. The portal is where you can schedule or reschedule 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, **call InformedDNA at 1-888-744-8952** to make alternative arrangements.

InformedDNA is the authority on the appropriate use of genetic testing. We counsel and advise health plans, health systems, and physicians. In addition, we provide clinical services, offering patients convenient access to certified genetic counselors.



4 Meet with your genetic counselor

At the scheduled time, your genetic counselor will call you to go over your test results 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 your test results



Discuss options to manage your health



Review your family health history to assess the risks to others in the family



Discuss potential reasons to speak with family members who may be at risk of having XLH

Counseling by:



Sponsored by:



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 patients or caregivers of patients aged 1 year and older who have been diagnosed with XLH.

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? **Call InformedDNA at 1-888-744-8952.**