X-Linked Hypophosphatemia (XLH)
A hereditary, progressive, and lifelong disease

What That Could Mean for You and Your Family

- Understand XLH: Symptoms and cause
- Learn the genetics of XLH: How it can be inherited
- Explore how to manage XLH: Today and tomorrow

Use this brochure to help you:

This information is for educational purposes only and is not intended to provide medical advice. Your provider should always be your primary source of medical advice for any health, diagnosis, or treatment practices.
What are some of the common symptoms of XLH?

Symptoms can differ for everyone and may vary among children and adults.

**Rickets** is a key feature of XLH in children and causes symptoms such as bowed legs, which become noticeable when the legs begin to bear weight.

**CHILDREN**
- Delayed and disproportionate growth
- Rickets

**CHILDREN AND ADULTS**
- Dental issues, including tooth abscesses (infection)
- Osteomalacia (weakening of bones)
- Bowed legs or knock-knees
- Bone and joint pain
- Short stature

**ADULTS**
- Enthesopathy (damage to the connective tissues between bones and tendons or ligaments, causing pain, swelling, and inflammation)
- Spinal stenosis (narrowing of the spaces within the spine)
- Osteoarthritis (inflammation of the joints)
- Recurring and low-trauma fractures

4 out of 5 adults with XLH have dental abscesses.

Learn more about the symptoms of XLH in adults at XLHLink.com.
What causes XLH?

When someone has XLH, their body makes too much of a protein called fibroblast growth factor 23 (FGF23). FGF23 plays a role in controlling the amount of phosphorus present in the body.

Too much FGF23 causes phosphorus wasting, which is the loss of phosphorus through the urine.

Phosphorus wasting leads to low levels of phosphorus in the blood (called hypophosphatemia), causing various symptoms of XLH.

Rachael and her children, Brooke and Benjamin, living with XLH.
XLH may also affect other members of your family

“I learned my diagnosis when I was 33 years old because I realized there was something not quite right with my little girl. Her legs were very bowed.

Once I found out XLH was the cause, it dawned on me—that went far beyond just my little girl. It was something that affected many members of our family, including myself.

We’re up to 21 living family members that have gotten a genetic test confirming their XLH diagnosis.”

Emily, living with XLH

“I always thought I had arthritis until I was diagnosed with XLH about a year ago.”

Allen, Emily’s uncle, living with XLH
XLH is inherited on the X chromosome*

The “X” in XLH stands for “X-linked” because the condition is due to a change (a mutation) on the X chromosome* that can be passed on to children. Males have XY chromosomes, and females have XX chromosomes.

This means that:

• Sons inherit an X from their mother and a Y from their father. Since XLH is carried on the X chromosome, a father cannot pass XLH on to his sons
• Daughters inherit an X from their mother and an X from their father, so they can inherit XLH from either parent

If a father has XLH, all of his daughters and no sons will have XLH.

If a mother has XLH, each of her children (boy or girl) has a 50% chance of inheriting XLH.

Shaded figures represent people affected by XLH.

XLH is not always inherited
Children can be born with XLH, even if there’s no family history of the disease. This is called a spontaneous case, and it can happen in 20% to 30% of people with XLH. If someone has a spontaneous case, they can still pass the disease on to their children.

*Chromosomes are thread-like structures made up of DNA that are found inside some cells of the body.
Confirming a diagnosis of XLH

Lab tests can help identify XLH

Fasting serum phosphorus
One important test measures the amount of phosphorus in the blood. When levels are low, this is called low serum phosphorus—a telltale sign of XLH.

Your doctor may also order blood and urine tests to check for other important signs, including levels of two forms of vitamin D and calcium. Both vitamin D and calcium are crucial components of good bone health.

Genetic and FGF23 tests
Your doctor may include a PHEX genetic test or an FGF23 protein test to help confirm a diagnosis of XLH.

Talk to your doctor for specific testing recommendations
The frequency of these tests and the ages at which they are performed vary widely based on a person’s symptoms, severity and progression of XLH, medical history, medications being taken, and more.

Monitoring XLH Over Time: What to Expect
Because XLH is rare and lifelong, it may be difficult to find information about what to expect as you age. This resource can help you understand important details about:

• XLH symptoms and progression
• Common tests for XLH
• Types of doctors you may encounter on your XLH care team
• Healthy living tips

Download the guide at XLHLink.com/Resources.
Find and work with a knowledgeable XLH care team

An XLH care team may look different for everyone since the type of specialist you see can vary. By staying informed and actively engaging with a healthcare team, patients can better understand how to manage XLH and become advocates for themselves and their affected family members.

Primary XLH Care Specialists

- Endocrinologist
- Geneticist
- Nephrologist
- Rheumatologist

Other Care Team Members

- Internal medicine
- Primary care provider
- Orthopedist / Orthopedic surgeon
- Occupational and physical therapists
- Dental specialist
- Pain specialist

Need an XLH specialist?

Visit XLHLink.com to find a specialist in your area.

Elizabeth and her son Simon, living with XLH
Get the support you need

Sign up at XLHLink.com to receive newsletters, additional resources, and information about managing XLH—and discover more from these online resources.

The XLH Network
XLHNetwork.org

Global Genes
GlobalGenes.org

National Organization for Rare Disorders (NORD)
RareDiseases.org

NIH Genetic and Rare Diseases (GARD)
RareDiseases.info.nih.gov

Register for educational events at XLHEvents.com. You’ll learn about XLH from clinical specialists and hear patients and caregivers share their stories of living with XLH.

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Todd and Heidi, with their daughter Ruby, living with XLH

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