YOUR JOURNEY WITH XLH

Shannon
Living with XLH

Explore facts about this rare disease. Start the conversation with your doctor today, and learn how to manage it.

XLH LINK | XLHLink.com

WHAT DOES XLH MEAN?

XL

The X and L stand for X-linked, which means that the condition is passed down through the X chromosome.

H

H stands for hypophosphatemia, a condition caused by low levels of phosphorus in the blood.

Jason and Alice, father and daughter who are living with XLH, and Alice's mother, **Lisa**

NOT SURE WHERE TO START?

No matter where you are in your journey with XLH, this brochure can help answer some of the questions you may have about this rare condition.

How to get diagnosed

Do you think you have XLH? These sections can help you learn more about XLH and how to get diagnosed:

- About XLH, pages 4-7
- Symptoms of XLH, pages 8-10
- How XLH is diagnosed, page 11

How to manage XLH

Have you been diagnosed with XLH and want to find out how to manage your symptoms? These sections can help you learn more about existing healthcare and support options:

- How symptoms can progress, pages 8-10
- Types of doctors who can help, pages 12-13
- Managing XLH, pages 14-15

Talk to your doctor to learn more about XLH and get answers to any questions you may have after reviewing this brochure.

XLH LINK

XLH IS GENETIC, LIFELONG, AND PROGRESSIVE

Symptoms can start early in life, and new ones may appear as you get older. XLH is not life-threatening, but symptoms can change or get worse over time

People with XLH have lower than normal levels of phosphorus in their blood. Phosphorus plays a key role in our bodies. It is found in our blood, muscles, and bones and helps with:

- **Strengthening bones and teeth**
- **Muscle and nerve function**
- **Energy levels**
- **Growth**

KaraBeth
Living with XLH

What causes low levels of phosphorus in XLH?

XLH is caused by a change (variant) in a gene called the *PHEX* gene. This variant causes the body to produce too much of a hormone called fibroblast growth factor 23 (FGF23).

The body produces too much FGF23

Extra FGF23 causes too much phosphorus to be lost through the urine instead of staying in the blood

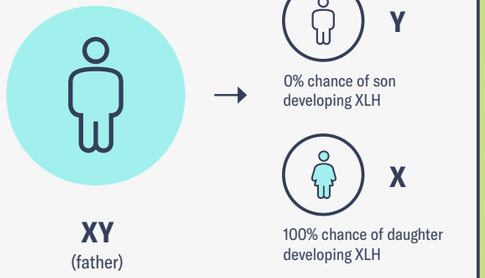
Low levels of phosphorus can lead to weakened bones, muscles, and teeth

Myth: XLH is just a vitamin D deficiency

Fact: XLH is not caused by low levels of vitamin D. It is a genetic disease (something you are born with) that leads to low levels of phosphorus in the blood.

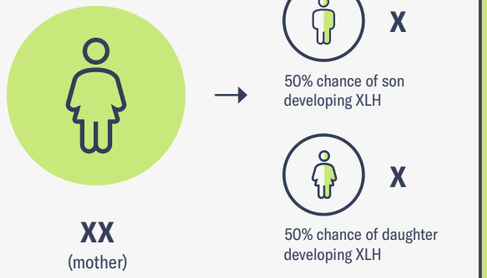
XLH CAN BE INHERITED OR HAPPEN SPONTANEOUSLY

XLH can be passed down through a variant in the *PHEX* gene, on the X chromosome. Chromosomes are structures inside your body's cells that contain your genes. Everyone has at least one X chromosome. Men have an X and a Y chromosome (XY), and women have two X chromosomes (XX).



If a father has XLH:

All daughters will have XLH. This is because **daughters inherit an X chromosome from their father**. No sons will have XLH because **sons get the X chromosome from their mother**.



If a mother has XLH:

Both sons and daughters have a 50% chance of having XLH. This is because **all children inherit an X chromosome from their mother**.

XLH does not always have a family history

XLH can also occur randomly. This is known as a spontaneous case of XLH. This means it is not passed down from a parent. Up to 3 out of 10 people have XLH because of a spontaneous gene variant. **People with a spontaneous case of XLH can pass it on to their children.**

Tracking XLH in your family

Because XLH is genetic, other people in your family might have it. One way to check is by filling out a family tree that shows who has XLH or signs of it. This is called a pedigree. Helping other family members find out if they have XLH can be the first step to getting the care they need.

KaraBeth, who is living with XLH, and her mother, **JoBeth**

> XLH has impacted our entire family. As parents, we spend a lot of time and energy on KaraBeth's healthcare.
>
> *JoBeth, caregiver*

A patient living with XLH

Myth: If XLH symptoms are mild, there's no need to talk to a doctor

Fact: XLH is a lifelong disease, and its symptoms can change or get worse over time. You don't have to wait for your symptoms to be severe to talk to your doctor about managing XLH.

THE SYMPTOMS OF XLH CAN CHANGE OR GET WORSE OVER TIME

One of the early effects of XLH is that it can make growing bones weaker. Over time, other symptoms can develop.

Symptoms that can begin early in life

BONES, MUSCLES, AND JOINTS

- Weakening of growing bones (rickets)
- Weakening of mature bone (osteomalacia)
- Delayed walking or problems with walking or keeping balance (gait abnormalities)
- Shorter than average height
- Bone and joint pain
- Muscle pain and weakness
- Bowed legs and knock knees
- Unusual shape of the head (craniosynostosis)

DENTAL

- Dental issues such as abscesses and tooth loss

OTHER

- Fatigue
- Early hearing loss

Symptoms that can develop later in life

In addition to symptoms that can appear early in life, symptoms may get worse or new symptoms can develop as you get older. These new symptoms may include:

BONES, MUSCLES, AND JOINTS

- Joint damage (osteoarthritis)
- Broken bones (fractures) and areas of weakened bone that are not completely broken (pseudofractures)
- Hardening of tissue attaching bones and muscles (enthesopathy)
- Narrowing of spaces in the spine (spinal stenosis)

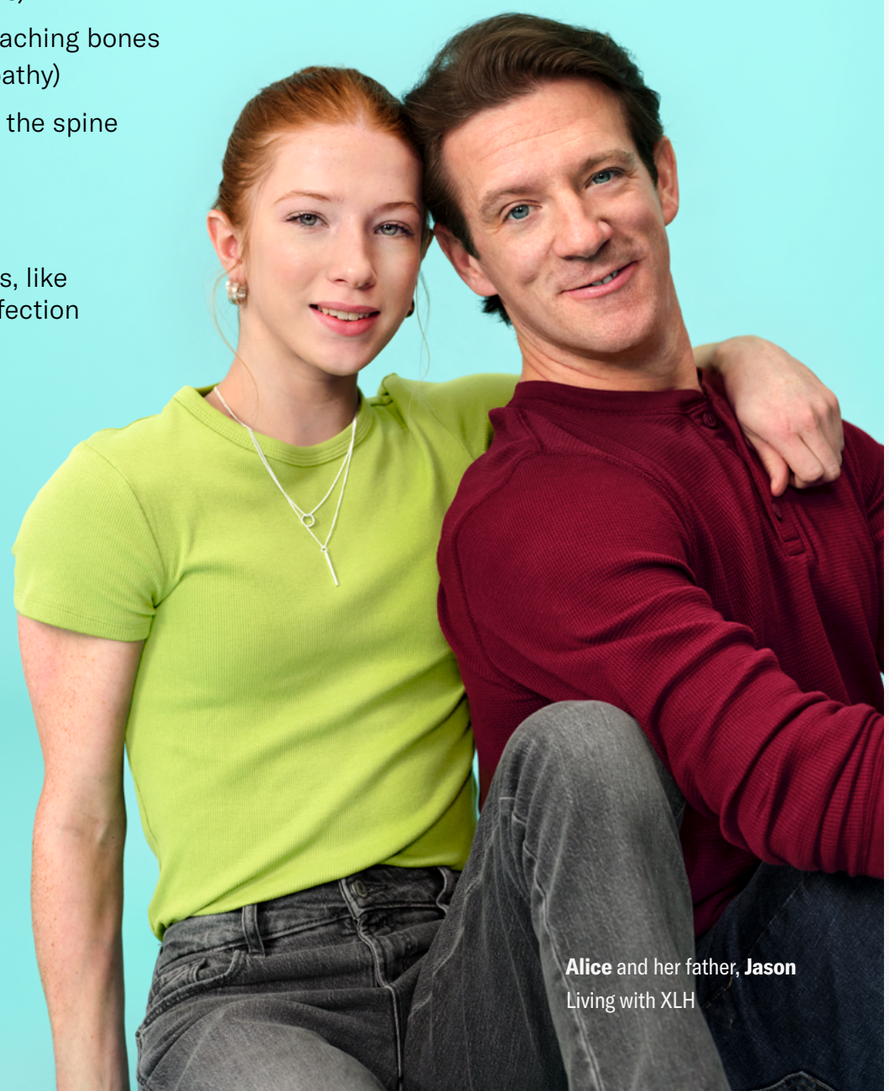
DENTAL

- Problems with the gums, like redness, swelling, or infection (periodontitis)

OTHER

- Hearing loss

Be sure to tell your doctor about all your symptoms so they can make a care plan that's right for you.



Alice and her father, **Jason**
Living with XLH

HOW XLH IS DIAGNOSED

XLH is a progressive disease, so it is important to get an accurate diagnosis to start a management plan with your doctor.

A family history can help confirm a diagnosis

Because XLH is a hereditary condition, your doctor may start by asking you about other family members. The goal is to find out if any of your relatives have XLH or similar symptoms to yours. In addition to looking for a family history of XLH, doctors can use a few different ways to diagnose XLH, like:

Signs and symptoms

X-rays

Blood tests for phosphorus levels

Genetic testing for a variant in the *PHEX* gene that causes XLH

Testing for XLH involves several steps. You may need to see more than one doctor to get an accurate diagnosis.

DOCTORS WHO CAN HELP DIAGNOSE AND MANAGE XLH

XLH is rare, so **it helps to find a doctor who is knowledgeable** about the condition. Your doctor (primary care provider) will likely play a role in helping you manage your XLH. But because the symptoms of XLH vary, **it may take different types of doctors to diagnose and manage it**.

Myth: XLH only affects children, not adults

Fact: Many people think that XLH only needs to be managed during childhood while bones are still growing, but this isn't true. XLH is a lifelong condition that can also impact adults.

KaraBeth and Alice
Living with XLH

Types of specialists who can diagnose and manage XLH

- **Endocrinologists** treat disorders that affect hormones like FGF23
- **Rheumatologists** treat disorders that affect joints, muscles, and connective tissue
- **Nephrologists** treat disorders that affect the kidneys
- **Geneticists** diagnose and manage genetic disorders

Additional specialists that can help

- **Orthopedists** treat bone and joint problems with orthopedic surgeons operating on damaged bones and joints
- **Dentists**
- **Physical therapists** help with movement issues
- **Pain specialists** help people manage bone, joint, and muscle pain
- **Ear, nose, and throat (ENT) doctors** diagnose and manage hearing loss

Need help finding a doctor who has experience with XLH?
Find a doctor near you.

EARLY MANAGEMENT OF XLH IS IMPORTANT

Even if the symptoms seem mild, waiting to manage XLH may lead to symptoms getting worse. Because XLH affects people their whole lives, it's important to stay consistent with care as you get older.

Raquel, who is living with XLH, and her sister, **Anne**

Treatment options

Possible treatment options that can help manage some of the symptoms of XLH include:

- **Medications and supplements**
- **Corrective bone surgeries**
- **Dental care**
- **Physical/occupational therapy and pain management techniques**

Talk with your doctor about the best ways to manage your XLH symptoms.

Learn more about ways to manage XLH and get support for children, teens, and adults.

Myth: I have dealt with my XLH symptoms by myself for a long time. I can keep dealing with them on my own

Fact: XLH symptoms can get worse. It's important to talk with a doctor about starting to manage your symptoms sooner rather than later.

XLH MAY BE RARE, BUT YOU ARE NOT ALONE

There are advocacy organizations and resources to help support you in your journey

XLHNetwork.org
A worldwide community of XLH patients, parents, caregivers, and medical professionals.

RareDiseases.org
The National Organization for Rare Disorders is a patient advocacy organization dedicated to individuals with rare diseases and the groups that serve them.

GlobalGenes.org
A globally connected community committed to eliminating the challenges of rare diseases and providing information, resources, and connections to all communities affected by these rare diseases.

GotTransition.org
Got Transition is a national resource center that aims to help children and young adults switch from pediatric to adult healthcare.

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I am grateful to my healthcare team, and I am glad to have a plan for my XLH. I continue to find a balance between managing my XLH and living my life.

Sunindiya, living with XLH

Sunindiya
Living with XLH

MANAGING XLH STARTS WITH A CONVERSATION—TALK TO YOUR DOCTOR TODAY

KaraBeth, Jason, Alice, Sunindiya, Raquel, and Shannon
Living with XLH

Download the XLH doctor discussion guide for your next appointment.

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KYOWA KIRIN

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