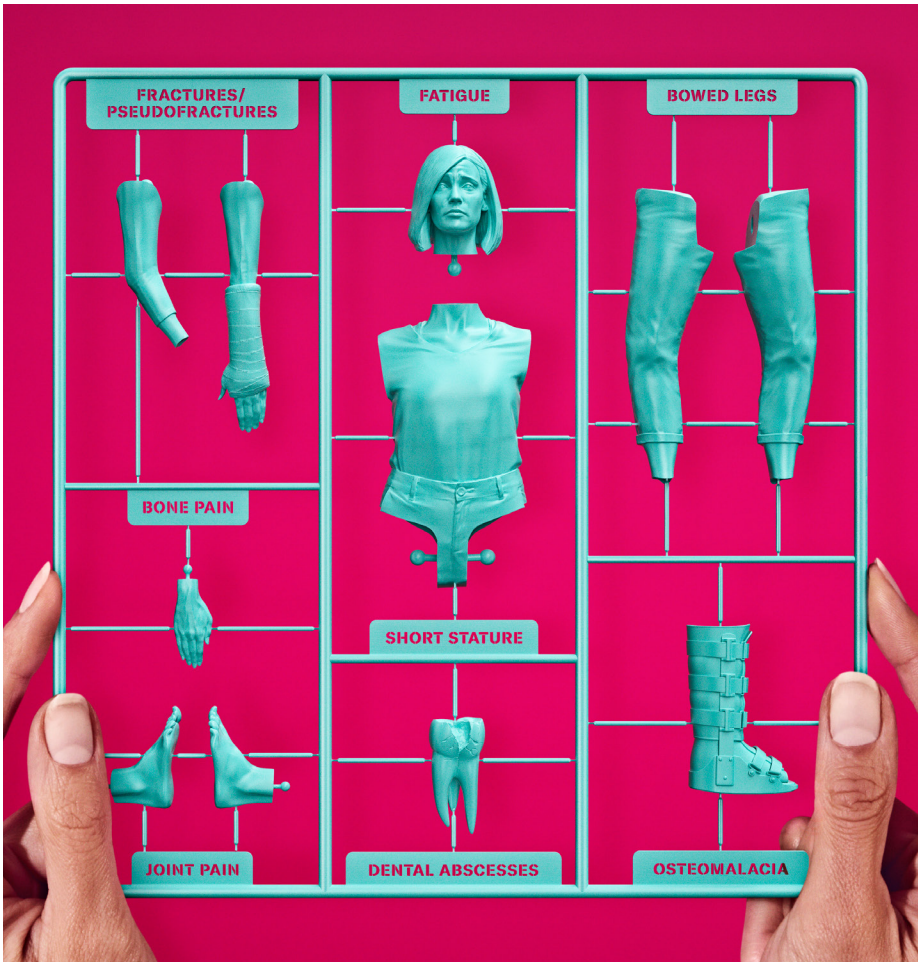
FOR PATIENTS

Are your symptoms more connected than you think?

It might be XLH

Talk to your doctor about X-linked hypophosphatemia (XLH)



XLHLink.com

XLH LINK

Table of contents

The basics of XLH

XLH is a rare and lifelong disease

XLH, or X-linked hypophosphatemia, is a hereditary, progressive disease that affects up to

1 IN 20,000 PEOPLE

What does XLH mean?

XLH

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

XL stands for X-linked, which means that the condition is most often passed down through the X chromosome.

3 Things to know about XLH

Hereditary

XLH is primarily an inherited condition, which means parents pass it down to their kids.

Progressive

New symptoms of XLH may appear at any age and can worsen over time.

Lifelong

People with XLH can continue to experience symptoms throughout their lives.

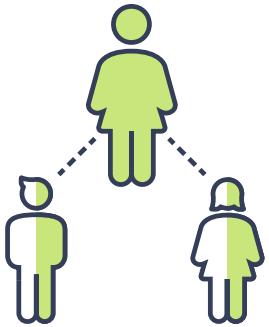
The cause of XLH

XLH is caused by a genetic mutation

A gene mutation causes the body to produce too much of a hormone called fibroblast growth factor 23 (FGF23):

Extra FGF23 makes the body release too much phosphorus through the urine

When phosphorus levels in the blood drop too low, this is known as hypophosphatemia, which can lead to weak bones

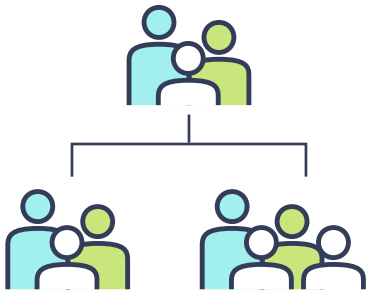


XLH is often passed down

You can inherit the condition from the X chromosome of either your father or mother.

XLH does not always have a family history

~20% to 30% of people develop XLH as the result of a spontaneous mutation. This means it's not passed down from a parent. They can, however, pass it on to their children.



The impact of XLH may go beyond you

If you have XLH, there's a good chance other family members may have it. Let relatives know about your diagnosis so they can find out if they have XLH.

WANT TO LEARN MORE ABOUT XLH?

Visit XLHLink.com for more information on how to support your XLH journey.

Symptoms of XLH

Most XLH symptoms start in childhood

The symptoms of XLH may seem unconnected. Some people think XLH is a disease that only affects children. The truth is, XLH continues into adulthood. Symptoms that started at an earlier age may worsen or change over time.

Existing symptoms from childhood may include:

Benjamin

Living with XLH

- Irregularities in the shape of the head
- Headaches
- Dental abscesses
- Tooth loss
- Rickets and osteomalacia*
- Delays or irregularities in walking
- Short stature
- Bowed legs and knock knees

*Rickets is a weakening of growing bone and osteomalacia is the weakening of mature bone, both due to low levels of vitamins and minerals, including phosphorus, calcium, and vitamin D.

Other symptoms:

- Muscle pain and weakness
- Bone and joint pain
- Fatigue

Some XLH symptoms start in adulthood

As XLH progresses, new problems may show up later in life that can decrease mobility and impact daily function.

Symptoms experienced in adulthood may include:

Rachael and her children, Brooke and Benjamin

Living with XLH

- Hearing loss
- Osteomalacia
- Hardening of ligaments or tendons
- Fractures and pseudofractures (areas of bone weakened by a lack of certain minerals, including phosphorus)

Other symptoms:

- Muscle pain and weakness
- Bone and joint pain
- Fatigue

THINK YOUR SYMPTOMS ARE XLH?

Talk to your doctor to learn more. An early and accurate diagnosis is important to manage this condition and help improve symptoms.

Actor portrayal

Diagnosing XLH

Medical tests

Your doctor may ask for some or all of these tests to accurately diagnose XLH:

- Blood samples to measure phosphorus levels
- X-rays to evaluate the condition of the bones

ASK FOR A PHOSPHORUS TEST

This test is not always included in standard blood tests, but it's a good way to diagnose low phosphorus levels in XLH. Your doctor may require you to fast before taking this test.

Family history

Because XLH is often hereditary, your doctor may recommend genetic testing and ask about other members of your family

Doctors who diagnose XLH

You may need to see a specialist

Because XLH is rare and the symptoms vary, it may take multiple doctors to correctly diagnose it and provide you with the personalized care you need. Some of these specialists may include:

Endocrinologists

Treat disorders that affect hormones

Rheumatologists

Treat disorders that affect the joints and muscles

Nephrologists

Treat disorders that affect the kidneys

Geneticists

Treat genetic disorders

XLH LINK

Diagnosing XLH isn't always easy

XLH is typically diagnosed during childhood, usually around 1-2 years of age, when a child begins walking. In some cases, however, a diagnosis may not be confirmed until adulthood.

Preparing for your doctor visits

Here are some strategies to help you make the most of your appointments:

- Be ready to share your medical and family history
- Write down any symptoms you have had as well as medications you're taking
- Bring a list of questions you may have and any educational resources that can help guide the conversation

SUSPECT XLH?

If you feel like your symptoms are more connected than you think, ask a specialist if it might be XLH. Find a specialist at XLHLink.com/specialist

Managing XLH

Comprehensive care is available

Your healthcare team will work with you to come up with a combination of XLH management options that work best for you.

In addition to the doctors mentioned on the previous page, your healthcare team may include physical and/or occupational therapists, dentists, and pain specialists.

Arlene and her daughter Gina
Both living with XLH

To help you manage and understand XLH, your team may suggest:

Medications and supplements

Ask your doctor about treatment options that may be right for you

Physical/occupational therapy and pain management techniques

These may help reduce pain and improve joint stability, flexibility, and muscle strength

Dental care

It's important to stay on top of any dental issues that may develop as a result of XLH

Genetic counseling

This can help you understand XLH inheritance patterns and identify other family members with the condition

XLH treatment options

Work with your doctor

There are treatment options available to help manage some symptoms of XLH. Please talk to your doctor to learn more about what is best for you.

Getting the conversation started

Before and during XLH treatment, it's important to discuss treatment goals with your doctor. These may include:

- Getting your phosphorus levels into a normal range
- Treating osteomalacia
- Treating and preventing pseudofractures and fractures
- Relieving bone pain and joint stiffness

HAVE YOU ALREADY BEEN DIAGNOSED WITH XLH?

Be sure to ask about managing XLH at your next appointment.

Just knowing that there was an answer and a reason why was probably the best part of learning my diagnosis."

EMILY AND HER DAUGHTER ISLA

LIVING WITH XLH

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Living with XLH?

These organizations provide support and advocacy for people and families living with rare diseases, including XLH:

XLHLink
XLHLink.com

The XLH Network
XLHnetwork.org

National Organization for Rare Disorders (NORD)
rarediseases.org

Global Genes
globalgenes.org