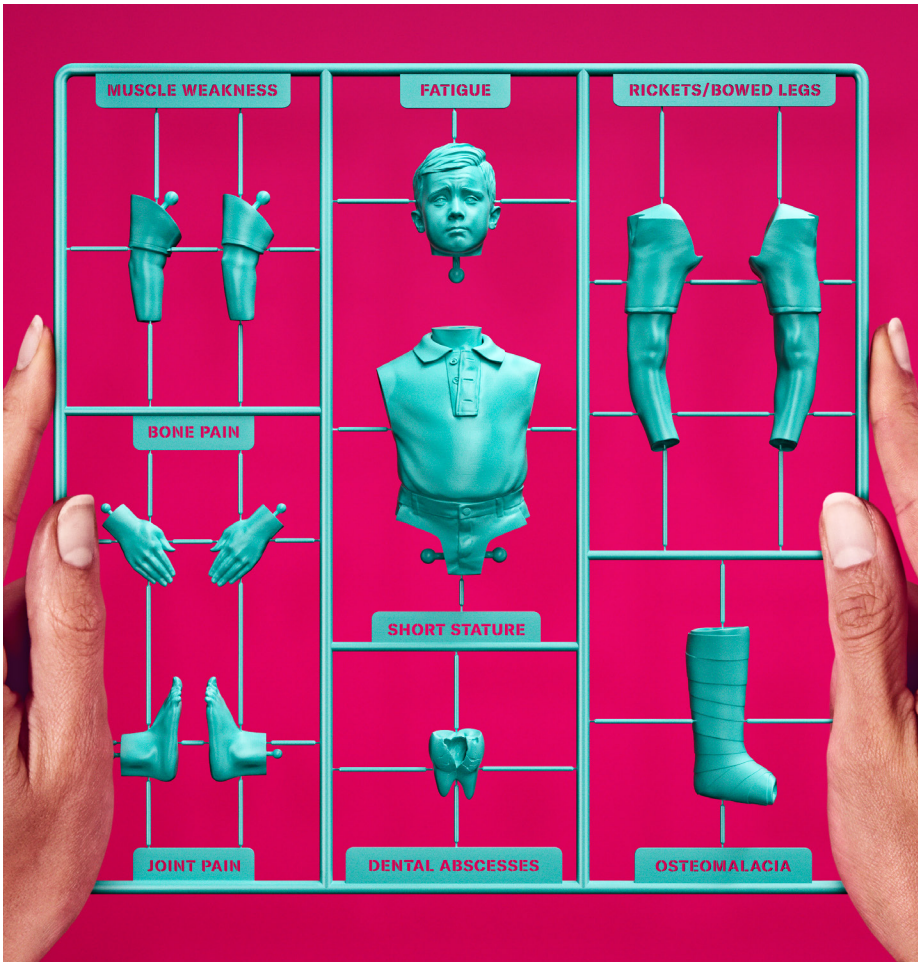
FOR CAREGIVERS

Are your child's symptoms more connected than you think?

It might be XLH

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



XLHLink.com

XLH LINK

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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

If your child has XLH, they will need your support throughout their journey.

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

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

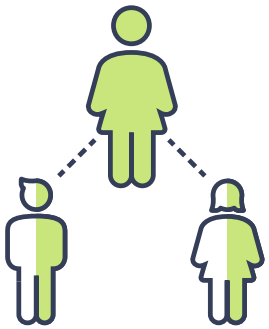
The cause of XLH

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

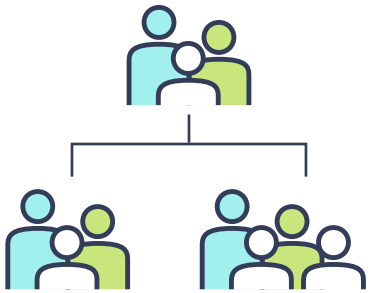


XLH is often passed down

Children can inherit the condition from the X chromosome of either their 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 your child

If your child has XLH, there's a good chance other family members may have it. Let relatives know about your child's 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 child.

Childhood symptoms of XLH

Are your child's symptoms connected?

Childhood symptoms 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

Other symptoms:

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

*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.

THINK YOUR CHILD'S SYMPTOMS ARE XLH?

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

XLH affects every child differently

Your child may not experience all the symptoms listed here. They may develop these or other symptoms later in life.

Some people think XLH is a disease that only affects children. The truth is, the symptoms of XLH can continue into adulthood, and they may change or worsen over time.

Elizabeth and her son Simon

Both living with XLH

Emily and her daughter Isla
Both living with XLH

Diagnosing XLH

Medical tests

Your child's 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 child's doctor may recommend genetic testing and ask about other members of your family

Doctors who diagnose XLH

Your child 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 your child with the personalized care they 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 child's doctor visits

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

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

SUSPECT YOUR CHILD HAS XLH?

If you feel your child's 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 your child.

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

Isla
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 your child

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 child's doctor

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

Getting the conversation started

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

- Getting your child's phosphorus levels into a normal range
- Treating rickets
- Addressing growth issues

HAS YOUR CHILD ALREADY BEEN DIAGNOSED WITH XLH?

Be sure to ask about managing XLH at your child's 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

XLH LINK

Caring for a child 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