XLH LINK

XLH AND YOUR FAMILY

XLH (X-linked hypophosphatemia) is a genetic condition that can be inherited, so it may affect other people in your family. Use this guide to understand XLH inheritance and support your family in taking the first step toward care.

XLH OFTEN RUNS IN THE FAMILY

XLH is a genetic condition. This means it can be passed from parents to their children through their genes.

“

My grandmother had XLH. She passed it on to my father, who passed it on to me.

Shannon
Living with XLH

XLH can also occur without a family history

This is called a spontaneous case, which occurs in up to 3 in 10 people living with XLH because of a new change in a gene.

People with a spontaneous case of XLH can still pass the condition to their children.

JoBeth
Karabeth’s mother

Karabeth
Living with XLH
(spontaneous)

THE SYMPTOMS OF XLH CAN VARY

Not everyone experiences XLH the same way. Some family members can have several symptoms, while others may only have a few. Even if XLH symptoms look or feel different, it's important to pay attention to all of them.

From left to right: **Karabeth, Alice, Jason, Sunindiya, Shannon,** and **Raquel.**
Alice and Shannon inherited XLH.
Karabeth, Jason, Sunindiya, and Raquel have spontaneous XLH.

There are several ways that XLH can affect the body

Do you recognize any of these symptoms in your family?

BONES, MUSCLES, AND JOINTS

- Bone and joint pain
- Shorter than average height
- Weakening of growing bones (rickets)
- Delayed walking or an unusual way of walking (gait abnormalities)
- Bowed legs and knock knees
- Muscle pain and weakness
- Weakening of mature bone (osteomalacia)
- Joint damage (osteoarthritis)
- Hardening of tissue attaching bones and muscles (enthesopathy)
- Broken bones (fractures) and areas of weakened bone that are not completely broken (pseudofractures)
- Unusual shape of the head (craniosynostosis)
- Narrowing of spaces in the spine (spinal stenosis)

DENTAL

- Issues such as abscesses and tooth loss
- Problems with the gums, like redness, swelling, or infection (periodontitis)

OTHER

- Hearing loss
- Fatigue

WHY THE "X" MATTERS IN XLH

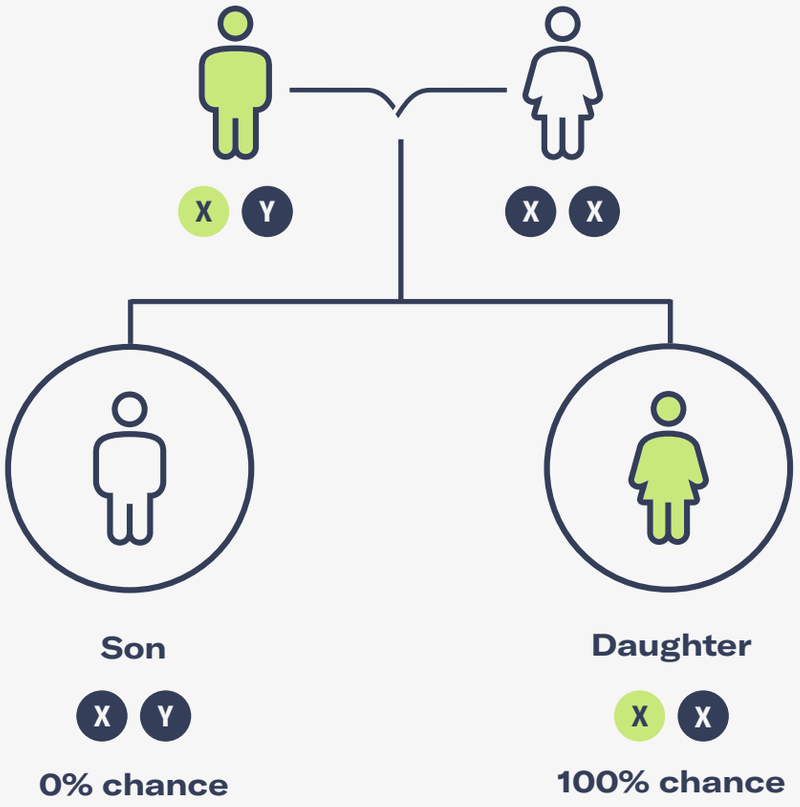
The X and L in XLH stand for X-linked, meaning the condition is passed down through the X chromosome

Everyone has at least one X chromosome:

- Men have an X and a Y chromosome (XY)
- Women have two X chromosomes (XX)

Chromosome with XLH

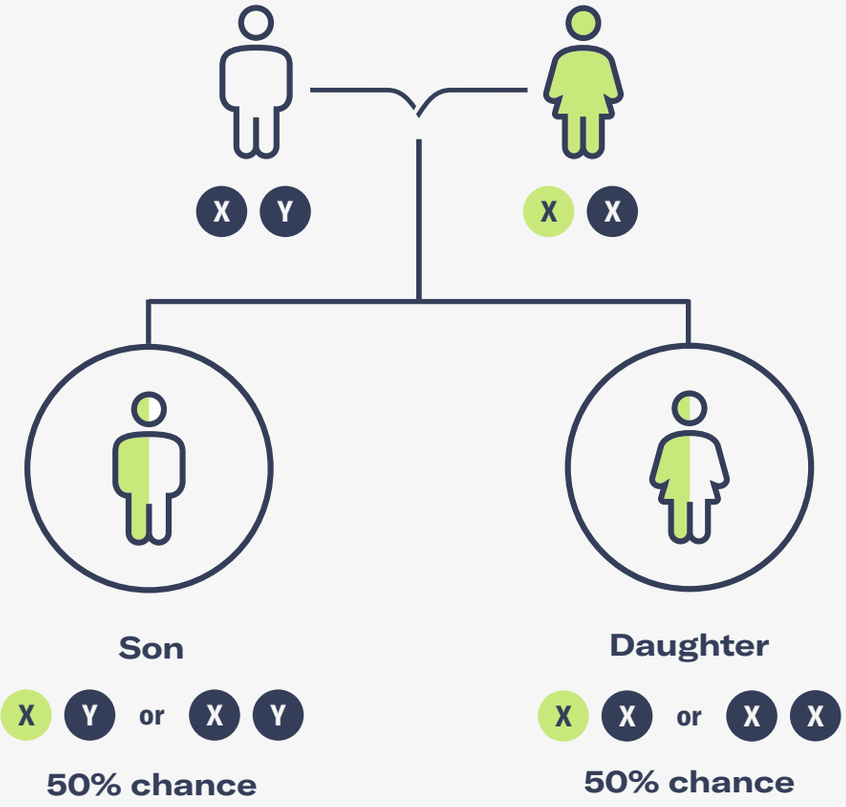
If a father has XLH and the mother does not:



All daughters will have XLH. No sons 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 and the father does not:



Both sons and daughters have a 50% chance of having XLH

This is because all children inherit an X chromosome from their mother.

XLH ACROSS GENERATIONS

Since XLH can come from a mother or a father, a family tree can show how XLH appears across different generations.

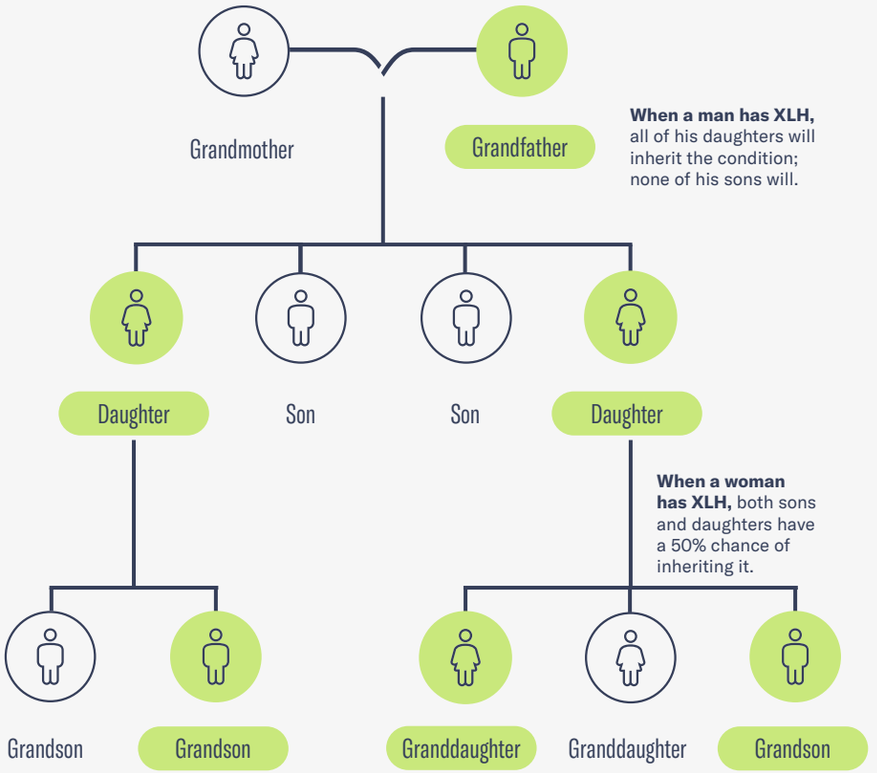
Alice and her father, **Jason**
Jason has spontaneous XLH.
Alice inherited XLH.

XLH family tree example

Below is one example of how XLH can be passed from a grandfather to some of his children and grandchildren.

To see how XLH may run in your family, use the worksheet at the end to mark up your own family tree.

Green indicates someone with XLH

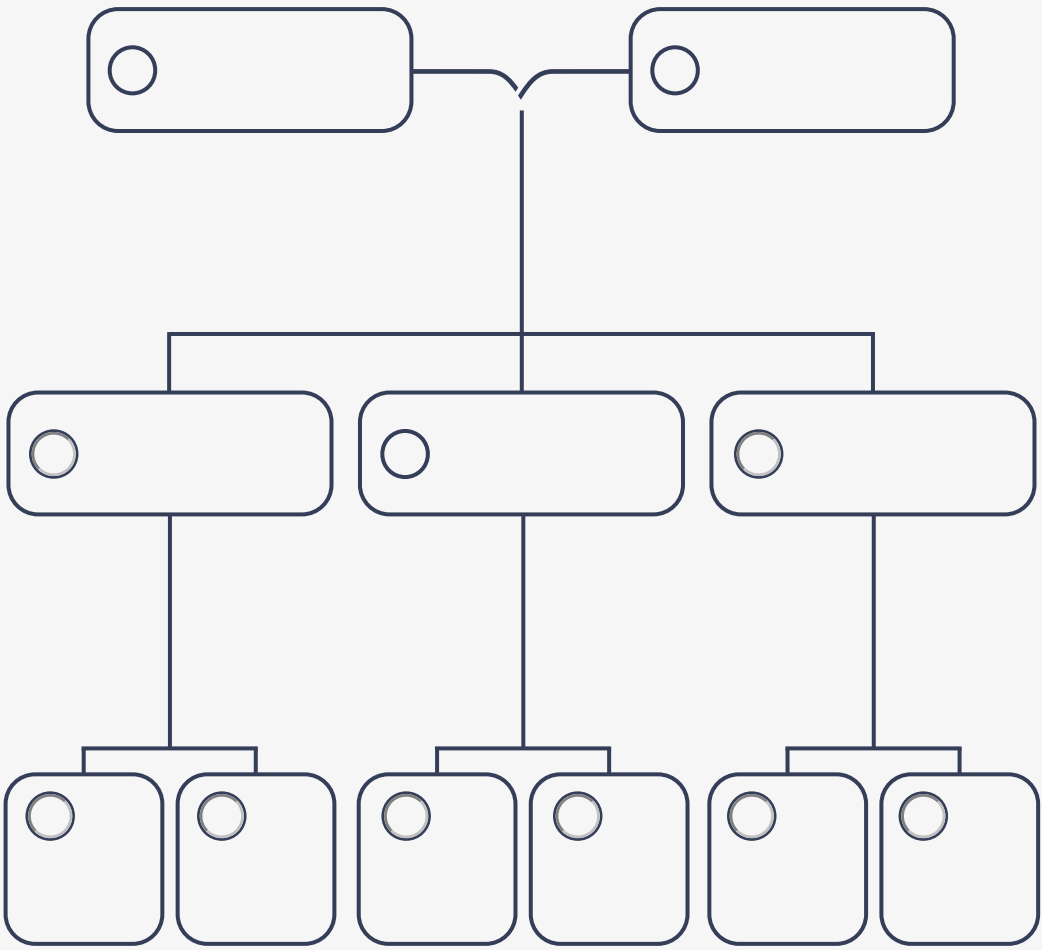


Reminder: Even when XLH occurs spontaneously, it can be passed along to children. A family conversation about inheritance is still important.

Your family tree

Using the example as a guide, create your own family tree

- Start by filling in the names of your family below
- Tear out this sheet along the dotted line and review the list of symptoms
- Mark the circle next to the name of any family member who has XLH or symptoms of XLH



Write down thoughts or questions on the back of this page and share with relatives. It can be a helpful tool for discussing how XLH runs in your family.

Notes

GET READY TO HAVE THE CONVERSATION

Although talking about health issues with a family member can be sensitive, starting a discussion around XLH could be the first step to getting them the care they need.

Consider these tips when you talk with your loved one:

Practice how to open the conversation

For example: "I've learned a lot about my condition lately, and it made me think about how it could be affecting other people in our family."

Share your experience with XLH

Some family members might feel nervous about talking to a doctor about their symptoms. Opening up about your XLH journey can help others feel empowered to start their own.

Offer them resources

Use the QR codes below to share a doctor discussion guide and a tool for finding an XLH specialist in their area. Information is also available at XLHLink.com. Let them know they don't need to make any decisions now—just that resources are available.

Be open to their response

They may be curious, unsure, or not ready to talk. That's okay! Let them know the door is always open if they have questions or want to talk again.

Help a family member talk with their doctor using our XLH Discussion Guide.

"My daughter's diagnosis also gave my husband the encouragement he needed to begin to manage his disease properly.

Lisa
Mother to Alice and wife to Jason, both living with XLH

Find a specialist.

CONNECT WITH OTHERS IN THE XLH COMMUNITY

XLH is rare, but you are not alone. Below are advocacy organizations and support groups you and your family can connect with.

The XLH Network

The XLH Network, a 501(c)(3) nonprofit organization, seeks to connect people around the world who are affected by or interested in learning more about XLH. The XLH Network connects affected individuals, families, and medical professionals.

National Organization for Rare Disorders (NORD)

NORD is a patient advocacy organization dedicated to helping individuals with rare diseases by driving advances in care, research, and policy.

Global Genes

Global Genes is a patient advocacy organization that works to build awareness, educate the global community, and provide resources to those affected by rare diseases.

Find a doctor near you with the Specialist Finder at XLHLink.com.

Visit us on social media

KYOWA KIRIN

Kyowa Kirin, Inc. is a proud sponsor of the XLH Network and NORD.

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