DIAGNOSED WITH XLH OR SUSPECT YOU MAY HAVE XLH?

Learn more about XLH, or
X-linked hypophosphataemia

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

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WHAT IS XLH?

XLH, or X-linked hypophosphataemia, is a **hereditary, progressive and lifelong disease** that can affect children and adults regardless of age.[1] It can have serious consequences in the bones, muscles and teeth.

Hereditary – XLH is an inherited disease, which means it runs in families. In some cases, XLH may occur spontaneously.

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

Lifelong – Individuals with XLH will continue to experience symptoms throughout their lives.

TAKE NOTE

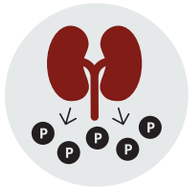
Doctors may call XLH by other names[2,3]:

- X-linked hypophosphataemic rickets
- Hereditary hypophosphataemic rickets
- Familial hypophosphataemic rickets
- Vitamin D–resistant rickets (VDRR)
- Vitamin D–resistant osteomalacia
- X-linked vitamin D–resistant rickets
- Hypophosphataemic rickets
- Hypophosphataemic vitamin D–resistant rickets (HPDR)
- X-linked rickets (XLR)
- Genetic rickets
- Familial hypophosphataemia

Benjamin, living with XLH

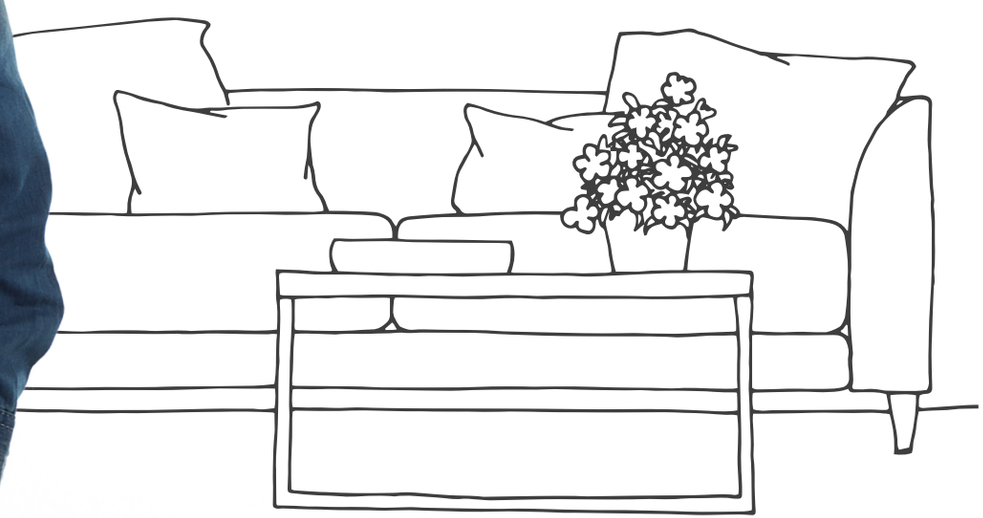
WHAT CAUSES THE SYMPTOMS OF XLH?[1,2,4]

People with XLH produce too much of a protein called fibroblast growth factor 23, or FGF23.



Too much FGF23 causes kidneys to lose phosphate through urine, leading to low levels of phosphate in the blood, called hypophosphataemia.

Chronic low phosphate causes bones to become weak and soft. Weak bones are the underlying cause of the symptoms of XLH.

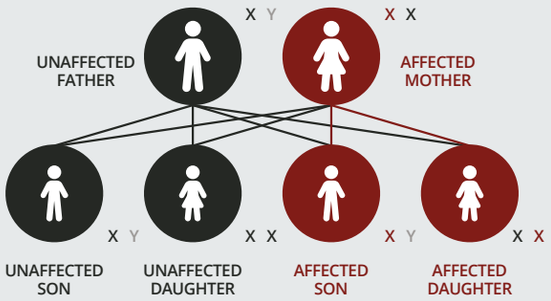


HOW DOES SOMEONE GET XLH?

XLH is inherited. The "X" in XLH stands for X-linked, because the disease is due to a defect (a mutation) in the X chromosome that can be passed on to offspring. Males have XY chromosomes, and females have XX chromosomes.[5]

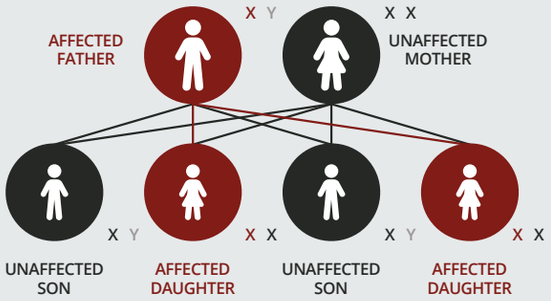
XLH INHERITANCE PATTERN

MOTHER WITH XLH



If a mother has XLH, each child has a **50% chance of inheriting XLH, regardless of sex.**

FATHER WITH XLH



If a father has XLH, **all of his daughters and none of his sons will inherit XLH.**

About 1/3 of XLH cases are **spontaneous**, which means that there is no family history of XLH. However, anyone who has XLH can pass it on to their children.

TAKE ACTION

Ask your doctor about genetic testing:

- If you or a relative has XLH, there's a good chance that other relatives have it too. If you suspect XLH, see page 8 for what to do next.
- As a hereditary disease, XLH impacts children and adults. Without management, it can lead to serious, painful, debilitating and permanent symptoms in adulthood.[6]

Elizabeth, her mother Jean and her son Simon, living with XLH

WHAT SIGNS AND SYMPTOMS INDICATE XLH?

In children[1,2]

- Rickets
- Delayed growth
- Delayed walking
- Craniosynostosis (unusual head shape)
- Headaches due to Chiari malformation (where brain tissue extends into the spinal canal)

In children and adults[1,2,3,6]

- Short stature
- Osteomalacia
- Bone and joint pain
- Wrists or knees that appear larger than normal
- Bowed legs or knock-knees
- Spontaneous tooth abscesses
- Muscle pain and weakness
- Joint stiffness
- Waddling gait

In adults[1,2,3,6]

- Fractures and pseudofractures
- Enthesopathy (hardening of tendons or ligaments)
- Osteoarthritis
- Spinal stenosis
- Dental complications
- Hearing loss
- Fatigue

Ben and his mom, Charlotte, living with XLH

TAKE ACTION

If you have experienced any of these symptoms, speak with a doctor who knows about XLH. See page 9 for more information about finding the right doctor.

HOW IS XLH DIAGNOSED?

Diagnosing XLH can be tricky because it is a rare disease. It is important to find doctors who are knowledgeable about XLH and can identify it.

A diagnosis of XLH is based on a combination of family history, symptoms and lab tests.[1,3]

Your doctor will take a **family history** by asking about family members and whether they have similar symptoms.

Bowed legs and knock-knees in a child is indicative of rickets. Doctors may order tests and X-rays and assess evidence of rickets using a rickets severity score (RSS).

Laboratory testing can confirm XLH, including tests that measure the amount of phosphate in the blood and urine.

Genetic testing can help confirm a diagnosis of hypophosphataemia.

XLH is typically diagnosed during childhood, usually within 2 years of age, when legs start supporting weight.[2]

TAKE ACTION

The right team of physicians and health care providers can make a difference in the care that you receive. **Pediatric and adult specialists who may diagnose and treat XLH include:**

- **Endocrinologists** – doctors who specialize in treating hormone conditions
- **Nephrologists** – doctors who specialize in treating kidney conditions
- **Medical geneticists** – doctors who specialize in treating genetic conditions
- **Orthopaedists** – doctors who specialize in treating bone conditions

Becky, living with XLH

HOW DO YOU MANAGE XLH?

Finding the right health care team that you are comfortable with is important. They can provide personalized care, support and disease education. Voice any concerns and ask questions about XLH to learn as much as you can.

Managing XLH is different for children and adults.

In children, treatment aims to:

- Address **growth issues**
- Correct **existing rickets** and **improve the mineralization of bones** and teeth
- Correct **leg abnormalities**
- Improve **physical function**

In adults, treatment aims to:

- Reduce the **pain, fatigue and stiffness** of the bones, muscles and joints
- Correct **leg abnormalities and fractures**
- Improve **bone and dental health**

The types of care you may need to properly manage your XLH may include[1-3]:

- Medications or dietary supplements
- Physical and occupational therapy
- Pain management
- Dental care
- Hearing consultation
- Genetic counselling

USEFUL WEBSITES FOR LEARNING TO MANAGE XLH

XLHLink.ca is an informational website for individuals, caregivers and families living with XLH that offers education on diagnosis, symptoms and disease management.

The Canadian XLH Network is a nonprofit organization serving the XLH patient community. Visit their site at **facebook.com/canadianxlhnetwork**.

TAKE ACTION

Children who have XLH will need to transition from pediatric to adult care as young adults. Parents and doctors can help them understand their disease and encourage them to take charge of their medical care.

Arlene, her son Jason and daughter Gina, living with XLH

HOW TO PREPARE FOR YOUR DOCTOR'S VISIT

If you are unsure whether you or a loved one has XLH, **talk to your doctor. Follow the checklist below to prepare.**

Make a list of symptoms.
Before your appointment, take a few minutes to write down all your current and past symptoms.

Know your family history.
Talk to your family and ask if anyone has been diagnosed with XLH or experienced symptoms. Take notes, discuss them at your appointment and ask about genetic testing.

Organize your medical history.
Keep a folder with health records such as test results, office visit notes and a list of current medications and any known allergies. Bring it with you.

Keep track of your questions.
Before the visit, write down any questions you may have. Take notes at each appointment so you remember important points.

Advocate for your health.
Take an active role and speak up when you have concerns and look for answers when issues arise.

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