X-linked hypophosphataemia (XLH) diagnosis

XLHLink

A guide for family members

Who is this leaflet for?

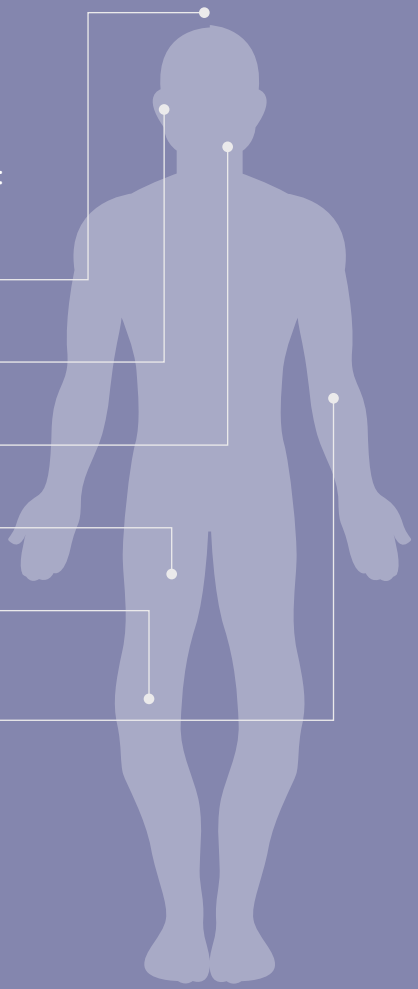
You have received this leaflet because someone in your family was diagnosed with X-linked hypophosphataemia, also known as XLH. Explore this leaflet to gain a better understanding of what this means to them and what it could mean for you. After reading through the information here you should have a better understanding of what XLH is, why you have received this leaflet, and whether you should speak to your healthcare provider about seeking a diagnosis yourself.

What is XLH?

XLH is a rare, lifelong genetic condition that affects about 1 in 20,000 people; its symptoms vary in children and adults.[1]

Some of the symptoms in adults and children include[1]:

- Short stature
- Hearing difficulties
- Tooth abscess
- Soft bones
- Leg bowing
- Pain in bones, muscle & joints



What causes XLH?

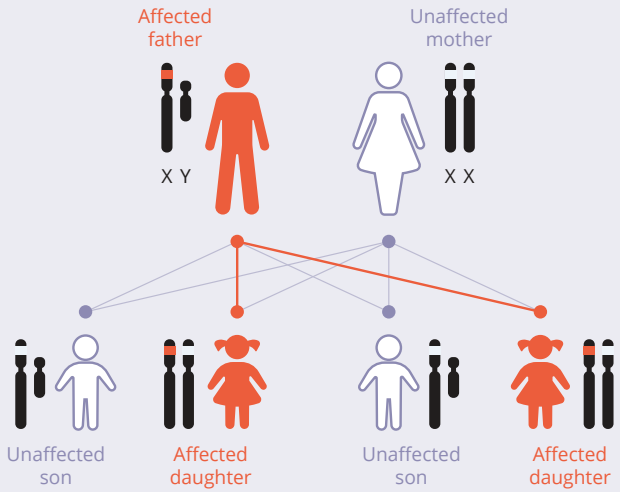
XLH is an inherited condition caused by a gene mutation on the X chromosome. This causes too much phosphate to be lost into the urine, and results in low phosphate levels in the body.[2] Phosphate is important for keeping bones, muscles and teeth healthy, so low levels can lead to soft bones.

While XLH can happen spontaneously, most people are born with XLH by inheriting an X chromosome with a gene mutation from one of their parents.[1]

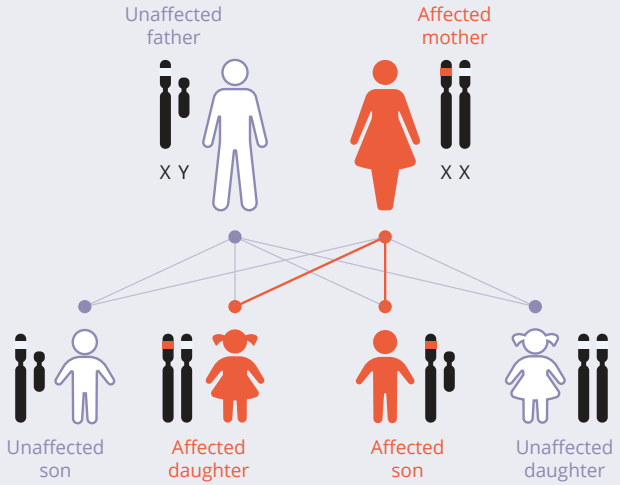
The X chromosome is one of the two sex chromosomes: women have two X chromosomes, while men have one X chromosome and one Y chromosome. Anyone with one X chromosome that has a mutated gene will have XLH.[1,2]

Because XLH is passed on through the X chromosome, mothers can pass it on to both sons and daughters; however, affected fathers pass it on only to daughters.[2]

XLH inheritance from an affected father



XLH inheritance from an affected mother



Who else in my family could have XLH?

Due to its inherited nature, in most cases, it is very important to investigate and trace XLH within the family to reach a diagnosis as early as possible for those affected.[3–5]

This process is known as family mapping, often referred to as pedigree analysis by healthcare professionals. You may have already spoken to your affected family member about this or it may be new to you. Either way, family mapping is a useful tool to identify members of your family who could be at risk and might benefit from getting a diagnosis. Take a look at the "What you need to know" leaflet with your affected family member to learn a bit more about the process of family mapping and how to make your own family map.

As the symptoms of XLH are progressive,[5] early diagnosis is important to protect the ones you love, especially children and younger family members. Early diagnosis allows timely interventions, which can help to reduce the impact of symptoms of XLH, including reduced growth in children and pain in adults.[4–5]

What are my next steps?

Talk to your healthcare provider. They can help you understand more about XLH, including whether family members are affected and the risk to future generations.[6]

Talk to your healthcare provider if you suspect you might have XLH. They can help you with diagnosis and management of XLH if you need it.

Talk to your family members to ensure that all your loved ones are aware of the condition and what decisions they might need to make. Drawing up a family map together can help you understand who might be at risk. See the "What you need to know" leaflet for more information or take a look at the resources below.

Helpful resources

Here are some resources you might find helpful. These can give you more information about the condition, the genetic risks, and other information that can help in talking to your healthcare provider and family about XLH.

- XLHLink.asia website **https://www.xlhlink.asia/**

References

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2. MedlinePlus. Hereditary hypophosphatemic rickets. Available at: https://medlineplus.gov/genetics/condition/hereditary-hypophosphatemic-rickets/. Accessed October 2020.
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4. Lambert A-S., et al. X-linked hypophosphatemia: Management and treatment prospects. Joint Bone Spine 2019;86(6):731-738.
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6. U.S. National Institute of Health. Genetic Mapping Fact Sheet. Available at: https://www.genome.gov/about-genomics/fact-sheets/Genetic-Mapping-Fact-Sheet Accessed June 2020.

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