INTERNATIONAL XLH ALLIANCE

X-LINKED HYPOPHOSPHATEMIA
A DISEASE FOR LIFE

WHAT IS XLH?

X-linked hypophosphatemia (XLH) is caused by a mutation of the PHEX gene causing the kidney to waste phosphate. This, in turn, leads to low levels of blood phosphate affecting the normal mineralisation of bones and teeth. Challenges begin in childhood when growth occurs and persist and often increase through adulthood.

XLH occurs in approximately 1 in 21-25,000 live births. It is inherited in an X-linked dominant pattern although 20-30% arise from spontaneous mutations. Along with family history, clinical manifestations and biochemical findings lead to a diagnosis of XLH.

Early Symptoms, like bowed legs or knocked-knees, become apparent when a child begins walking. XLH involves more than just bones and the impact on individuals and their families can be profound.

SYMPTOMS OF XLH

In addition to lower extremity abnormalities, some clinical manifestations of XLH in childhood include abnormal development of the skull, short stature, rickets, and dental abscesses. Bone and muscle pain are also common.

These symptoms may persist into adulthood with the addition of calcification of joints and tendons (enthesopathy), early onset osteoarthritis, spinal stenosis and hearing loss. Adults with XLH may suffer from fatigue, muscle weakness and are likely to have non-traumatic fractures and delayed fracture healing.

IMPACT OF LIVING WITH XLH

XLH is a lifelong disease. The physical and psychological complications affect children and adults.

XLH carries a stigma. The psychological challenges of living with a genetic disorder include knowing it can be passed to children. This is an important consideration that can cause emotional distress. Lifelong short stature carries the potential for being made fun of and bullying. XLHers may need adaptations at home, at work and at school. Depression may result from disability that impacts the ability to work and pursue typical daily activities.

WHAT CAN BE DONE TO IMPROVE OUTCOMES IN XLH?

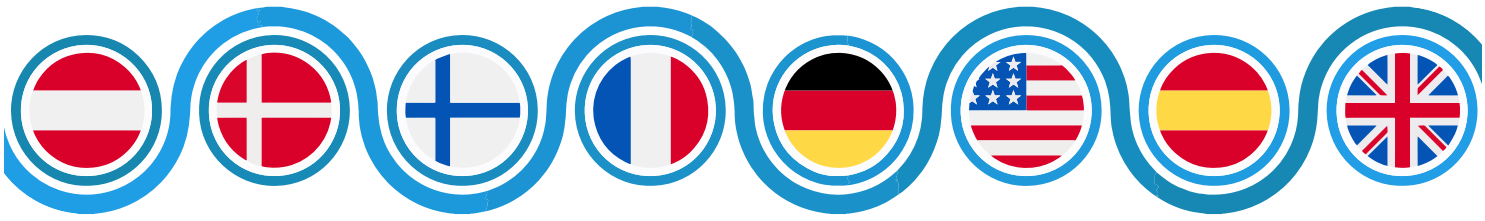
- The medical and care community, as well as payers, need to recognize that XLH is a life-long condition, which is incurable and brings a wide range of debilitating symptoms.
- Early and accurate diagnosis is needed so that treatment can be initiated promptly; all newborn babies where there is a family history of XLH should be tested for the relevant mutation.
- Primary care physicians should be educated about the disease and the need for urgent referral to a specialist.
- All patients should be assessed and managed by multidisciplinary care teams including medical and support services
- Timely and affordable access to medication based on individual need is important, with no differences based on country of residence or socioeconomic circumstances.
- Social services should be aware of the disease and its complications to ensure support for both the patient and their caregivers.
- International evidence-based treatment guidelines, developed with input from people living with XLH, are needed to ensure consistency across all affected people.
- Easily accessible, reliable disease information should be provided to patients to avoid assumptions or outdated knowledge.
- Education about patient rights should be provided to patients and caregivers to help them access the right treatment and support.
- Continued research on improved treatments should be conducted and supported.

WHERE TO GET MORE INFORMATION AND SUPPORT ABOUT XLH

The International XLH Alliance can put you in touch with an XLH organisation in your country (where one exists) that can provide information and support to people affected by the disease.

HOW YOU CAN HELP

- You can support people with XLH by sharing knowledge about the disease.
- You can contribute to fund-raising efforts for the organisations and for XLH research.
- You can volunteer your time to support your national group or the International XLH Alliance.



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