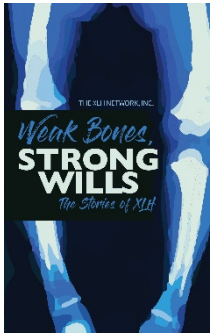
About The XLH Network, Inc.

The XLH Network, Inc., a 501(c)(3) nonprofit corporation, works to connect affected families, clinicians, and researchers around the world with up-to-date information on the diagnosis, symptoms, and treatment of hypophosphatemia in patients of all ages.

Our services include a moderated, peer-to-peer community forum; a physician referral database; educational literature and videos for patients and clinicians; a combined patient registry and natural history study; a speakers' bureau; and an annual patient education and networking event known as XLH Day.

Patients' Stories about Hypophosphatemia



There is much about living with XLH that is not in the scientific literature. *Weak Bones, Strong Wills: The Stories of XLH* contains inspiring stories by people affected by XLH. Not the medical jargon, not the numbers and charts, but the real-life experiences of patients and their families and friends. You can purchase your copy at your favorite online bookstore.

Scientific Literature on Hypophosphatemia

For an excellent overview of hypophosphatemia diagnosis and the prognosis for long-term health, check out "A Clinician's Guide to X-Linked Hypophosphatemia," in the *Journal of Bone and Mineral Research (2011).*

The Network's Mission

The mission of The XLH Network, Inc. is to:

- Promote awareness and education for affected families, medical professionals, and the community at-large;
- Support providers of medical care for better diagnosis and treatment;
- Create resources and a community for affected families so they can understand and cope with the complications of the disease;
- Foster the search for a cure.

If you would like to help us accomplish our mission, donations are tax-deductible and may be made online using the link at our website, XLHNetwork.org. Checks may be sent to The XLH Network, Inc., 911 Central Avenue #161, Albany, NY 12206.

XLH NETWORK

Connecting, Educating, Advocating

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About Hypophosphatemia

The XLH Network, Inc. represents the entire community of patients and families affected by genetic or tumor-based hypophosphatemia.

Hypophosphatemia refers to a low level of phosphorus in the blood. For our patient community, the hypophosphatemia is due to excessive levels of a hormone known as FGF23, which causes the kidneys to waste phosphorus.

Hypophosphatemia is a rare genetic disease, affecting approximately one in 20,000 people. It is passed from one generation to the next, although spontaneous genetic mutations may occur in a family with no previous history of the condition.

The most common version of this rare condition is X-linked Hypophosphatemia (XLH), with the affected gene carried on the X chromosome. It is one of the very few X-linked conditions inherited in a dominant manner, which means that only one parent needs to have the genetic mutation for the condition to be transmitted to the next generation. Approximately three hundred mutations have been identified as causing XLH.

There are also three other types of familial hypophosphatemia for which the genetic defect is not on the X chromosome. They are Autosomal Dominant Hypophosphatemia and Autosomal Recessive Hypophosphatemia Types 1 and 2.

Also, an ultra-rare condition known as Tumor-Induced Osteomalacia (TIO) is caused by a tumor that excretes excessive amounts of FGF23, and generally manifests in adulthood, although it can occur in childhood.

Diagnosis

Early diagnosis and life-long treatment are critical to the bone, dental, muscle, and emotional and social health of patients. A diagnosis can be made at any age and is usually based on the following biochemical tests:

- X-ray evidence of rickets/osteomalacia
- Low phosphorus levels in blood
- Normal calcium levels in blood
- Elevated alkaline phosphatase levels
- High phosphorus levels in urine
- Normal vitamin D levels in blood
- Normal or elevated serum FGF23 levels

Whole-Body Symptoms

XLH and the other genetic hypophosphatemias are life-long, whole-body disorders, affecting not just bones and teeth but also muscle function and energy levels. Symptoms range in severity and may include some or all of the following in the absence of life-long treatment:

- Lower limb deformities
- Short stature
- Waddling gait
- Spontaneous dental abscesses
- Bone pain in the absence of trauma
- Muscle pain and weakness
- Joint pain and early-onset arthritis, in the ankles, knees, hips, and spine
- Calcifications and bone spurs in the spine and joints
- Vertigo, hearing loss or tinnitus
- Extreme fatigue

Disclaimer

Information presented in this brochure is provided solely for educational purposes. All patients should consult their own doctors to get the best possible medical advice concerning their specific conditions and appropriate treatment.

Life-long Treatment

There are numerous misconceptions about how and when hypophosphatemia should be treated, and this is one of the reasons our organization continues to educate and advocate on behalf of our patient community.

Burosumab, a new treatment marketed as Crysvita®, has recently been approved by the U.S. Food & Drug Administration and the European Medicines Agency. While this treatment is not a cure, it is a major breakthrough which improves the kidney's reabsorption of phosphate by inhibiting the action of the FGF23. More research is needed to fully understand burosumab's long-term effects, but the experience to date in both children and adults indicates improvements in serum phosphorus and in bone mineralization, as well as reduced pain and fatigue..

An alternative approach which has been used for many years employs multiple daily doses of phosophorus and an activated form of vitamin D, calcitriol, essentially attempting to replace the urinary losses of phosphate. This treatment has required frequent monitoring in order to limit various complications including gastrointestinal distress and nephrocalcinosis (kidney calcification). Despite this therapy surgical correction of skeletal defects has been necessary in many treated subjects.

After six decades of research, there continue to be misconceptions about the disease. The XLH Network, Inc., is committed to connecting patients and doctors, educating them about XLH as a whole-life, whole-body disease, and advocating for additional research for our families.