Treatments

Currently, there is no cure for hypophosphatasia. In 2015, an enzyme replacement therapy for HPP by subcutaneous injections of asfotase alfa (Strensiq™) was approved essentially for pediatric-onset HPP. Other medications may be evaluated in clinical studies.

Patients and their families are advised to seek expert dental care and may benefit from physical therapy. Some people with HPP may be helped by a procedure in which load-sharing rods are inserted into the bone to prevent or heal fractures. Severely affected infants may have increased levels of calcium in their blood that may be treated with calcium-restricted diets. Doctors should avoid giving calcium supplements or vitamin D unless tests show a clear deficiency.

Because the prevalence of HPP is unknown, Soft Bones urges all patients to register with the International HPP Patient Contact Registry. This also allows patients and their families to stay informed of research opportunities and clinical trials.

For more information, contact Info@SoftBones.org

Soft Bones: Making a Difference

As a rare medical condition, HPP presents significant challenges to patients, their families, and caregivers. Finding a physician who can diagnose and treat HPP is often challenging because few doctors have HPP-specific training or experience. Soft Bones encourages patients and their doctors to become a team, to address the patient needs and alleviate complications. There is a need to bolster the education and training of HPP in the medical community—and to encourage medical staff to partner with patients in their treatment and care. Patients need more resources and support.

Soft Bones supports research, education, awareness, and policy and patient advocacy. We award research grants to scientists working to understand and treat HPP. We work with the NIH to advocate for government funding for research, and with government agencies to ensure patients receive their entitled benefits. Our role as a sounding board, advocate, and resource to the newly diagnosed patient and caregiver is most important.

How Can You Help?

Volunteer – we need volunteers to help Soft Bones fulfill its mission.

Fundraise – we always need enthusiastic and committed fundraisers. You can help sell bracelets, t-shirts or organize a fundraiser to show your support.

Donate – go online to www.SoftBones.org to donate today, or you can send a check to our address below. We are a 501c3 organization and your contribution is tax deductible.

For more information, please contact

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HYPOPHOSPHATASIA

Hypophosphatasia (hī-pō-ˌfäs-fə-ˈtā-zh(ē-)ə

is an inherited (genetic) metabolic disorder. People with the condition have low levels of the enzyme alkaline phosphatase (ALP), which impairs the mineralization of their bones. Normal mineralization is essential for hard and strong bones. Without it, bones and teeth become weak and soften, often causing skeletal deformities, fractures, premature tooth loss, and pain.

Soft Bones Foundation was formed in 2009 to provide information and a community to educate, empower and connect patients living with hypophosphatasia (HPP), their families and caregivers. The Foundation also promotes research of this rare bone disease through awareness and fundraising efforts.

Signs and Symptoms of HPP

The signs and symptoms of HPP vary widely in severity ranging from very mild to severe and sometimes life-threatening. Typically, the earlier HPP is diagnosed, the more severe the condition.

A hallmark of the disease is the deficiency of calcification of the skull and other bones. For infants, skull deformities can develop and can cause pressure on the brain, known as craniosynostosis. HPP manifesting in infancy or childhood is often misdiagnosed as it can resemble nutritional rickets (a deficiency of vitamin D) on X-rays.

Generally, bones and teeth are most often affected. Bones can become soft or weakened, which may result in fractures. Baby teeth can fall out earlier than normal, and fall out with the root still present.

Doctors typically classify HPP according to the age at which symptoms first appear: perinatal, infantile, childhood, adult, or odonto forms. Researchers understand HPP is a genetic condition of the TNSALP gene, which is part of a patient's genetic makeup, regardless of when it is diagnosed. It is clear that there is a range of symptoms and the level of severity of disease appears to correlate with the age at which patients first become symptomatic.

Forms of HPP

Perinatal

- In perinatal HPP, severe disease is apparent at birth.
- Typically, there are short limbs, abnormal chest shape and soft skull bones.
- This is the most severe type of HPP, with life-threatening complications. Parents who suspect their children to have perinatal HPP can consult an HPP specialist for detailed and expert evaluation.

Infantile

- Infantile HPP is diagnosed after birth, but before six months-of-age.
- HPP in infants is often more severe than it is in older children, with an estimated one of two such infants dying from the disease.
- Bones can become weak and soft from rickets leading to skeletal deformities.
- Infants with HPP can have difficulty gaining weight and have problems eating and breathing. They also have too much calcium in their blood. This can cause vomiting and kidney problems.

Childhood

- Childhood HPP is typically less severe than HPP in infants.
- Baby teeth typically fall out earlier than normal, often one of the first signs of HPP.
- Legs may be bowed or "knock-kneed," wrist or ankle joints may be enlarged, and the skull may not be shaped normally.
- Children with childhood HPP may also be weakened and experience a delay in gross motor skills such as sitting, crawling, or walking.

Adult

- Typically appears in middle age, but is often incorrectly diagnosed as osteoporosis.
- Fractures often reoccur in the feet and heal slowly.
- Adult bones can become soft (osteomalacia).
- Adults with HPP sometimes remember having rickets.
- There may be an increased risk for joint pain and swelling.

Odonto

- Only apparent clinical abnormality is dental disease.
- Mildest form of HPP.

"The truth is there is still a lot we don't know about HPP. It's an area where we still have much to learn. However, by coming together, sharing experiences, supporting research, and uniting our efforts, we can have a stronger voice and move closer toward a cure."

— Deborah Nettune Fowler, Soft Bones Founder