TREATMENTS

"Up until recently management of HPP has been purely supportive with symptom management and orthopedic treatment. Recently, based on promising results of clinical trials to date and a good safety profile, Health Canada has issued a Notice of Compliance with Conditions for asfotase alfa, human recombinant bone-targeted alkaline phosphatase for the treatment of confirmed pediatric-onset HPP. This is a very promising advance for the treatment of this disorder."

-Dr. Cheryl Rockman-Greenberg

MDCM, FRCPC, FCCMG Program in Genetics and Metabolism, WRHA

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 fractures. Severely affected infants may have increased levels of calcium in their blood that may be treated with calcitonin and certain diuretics. Doctors should avoid giving calcium supplements or vitamin D unless tests show a clear deficiency.

It has been estimated that severe forms of HPP occur in approximately one per 100,000 live births. The more mild childhood and adult forms are probably somewhat more common. On average one out of every 200 individuals is a carrier for HPP, but in certain Mennonite populations in Canada the carrier rate increases to one out of 25.

Soft Bones Canada - 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 specific training or experience. Soft Bones Canada encourages patients and their doctors to become a team, with the aim to address the patient needs and alleviate symptoms. There is a need to bolster education and training of HPP in the medical community and to encourage medical staff to partner with patients in their treatment and care. Patients also need more resources and support.

Soft Bones Canada supports research, education, awareness, policy and patient advocacy. Our mandate is to award research grants to scientists working to understand and treat HPP. Our Scientific Advisory Board is dedicated to seeing that the best care is available to patients across Canada. Our most important role is that of a sounding board, advocate and resource to the newly diagnosed patient and caregiver.

How Can You Help?

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

Donate - your donations help us support patients and families, distribute educational resources, and fund research. Donations can be made on our website: www.softbonescanada.ca

Spread the Word - tell people what you know about hypophosphatasia, or direct them to our resources to access more information.

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Soft Bones Canada

Hypophosphatasia

Hypophosphatasia (hypo-fahs-fuh-TAY-shuh) is a potentially lethal inherited metabolic disorder. People with the condition have low levels of the chemical alkaline phosphatase, which affects the mineralization of their bones. Normal mineralization is essential for hard and strong bones. Without it, bones and teeth become weak and soften, causing skeletal deformities, fractures, premature tooth loss and pain.

Soft Bones Canada was formed in 2013 to provide information and a community to educate, empower, and connect patients living with hypophosphatasia (HPP), their families, and caregivers.

Soft Bones Canada also promotes research of this rare bone disease through awareness, advocacy, and provision of research grants.

SIGNS AND SYMPTOMS OF HPP

The signs and symptoms of HPP vary widely, with symptoms from mild to severe. Typically, the earlier the condition is diagnosed, the more severe the condition. HPP discovered in utero, or in the first few days of life, was often fatal.

A hallmark of the disease is the lack of calcification of the skull and other bones. For infants, skull deformities can form and can cause pressure on the brain, known as craniosynostosis. HPP diagnosed at infancy and childhood is often misdiagnosed as it closely resembles 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 intact.

In the past, doctors classified HPP based on the age of the person at the time of diagnosis: perinatal, infant, childhood and adult. Now researchers understand HPP is a genetic condition of the ALPL gene, which is part of a patient's genetic makeup, regardless of when it is diagnosed.

While doctors are moving away from classifying the disease in different forms, it is clear that there is a range of symptoms and the level of severity of disease appears to correlate with the age that patients become symptomatic.

HPP ACROSS THE AGE SPECTRUM

Perinatal (before, during, and after birth)

» In perinatal HPP, the disease is apparent prior to birth and may be seen with an ultrasound.

» Infants with HPP are born with short limbs, abnormal chest shape and soft skull bones.

» This is the most severe type of HPP, with potentially life-threatening complications. Parents who suspect their children have perinatal HPP should consult an HPP specialist for detailed and expert evaluation.

Infant

» Infantile HPP is typically diagnosed shortly after birth.

» HPP in infants is often more severe than it is in older children, with one in two infants dying from the disease.

» Bones can become weak and soft leading to rickets and other 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 onset of HPP is often less severe than HPP in infants.

» Baby teeth typically fall out earlier than normal, often one of the first signs of HPP.

» Legs may appear 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 experience a delay in gross motor skills such as sitting, crawling or walking.

Adult

» Typically appears in middle age but is often misdiagnosed 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.

» Adults may be of short stature, with bowed or deformed legs.

"The regulatory approval of asfotase alfa represents an important milestone. Until now, there was no therapeutic option for patients. Yet there is still much work to be done. SBC remains committed to ensure patients have access to this important therapy by uniting our efforts to support ongoing research, to learn more about HPP, and how to effectively manage it. Our hope is that the awareness raised around the approval of this treatment will help more patients obtain an accurate diagnosis, and ultimately have better outcomes and an improved quality of life."

-Debbie Taillefer, Founder Soft Bones Canada