What Is Hypophosphatasia?

Hypophosphatasia (hī-pō-ˌfäs-fə-ˈtā-zh(ē-)ə) is an inherited (genetic) ultra-rare, metabolic (chemical) bone disease of broad-ranging severity that causes life-threatening disease in approximately one per 100,000 live births. People with the condition have low levels of the enzyme alkaline phosphatase (ALP), which impairs the mineralization of bones.

Normal mineralization is essential for hard and strong bones. Without it, bones become weak and soften and teeth may fall out prematurely (prior to 5 years of age). Depending on the severity of the skeletal disease, symptoms can include deformity of the limbs and chest, pneumonia, recurrent fractures, premature tooth loss and pain.

What are the signs and symptoms of hypophosphatasia?

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.

Symptoms of HPP May Include

- Short limbs, abnormal chest shape and soft skull bones
- Skull abnormalities such as craniosynostosis
- Calcified kidneys
- Failure to thrive of infants with difficulty gaining weight, eating, delayed gross motor skills and breathing
- Early tooth loss (before age 5)
- Soft bones prone to fracture; bowed long bones or “knocked-knees”, wrist or ankle abnormalities
- Pain including joint pain and swelling, often misdiagnosed as fibromyalgia.

Doctors typically classify HPP based on the age of the person at the time of diagnosis: *perinatal, infantile, childhood and adult* 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.

There is still a lot to learn about the role of alkaline phosphatase in the body. Many patients are misdiagnosed with rickets (nutritional or x-linked hypophosphatemic), fibromyalgia or osteoporosis. There are also considerable questions around the role of ALP on the body’s neurochemistry. More research is needed in this area to understand the full implications.

What is the treatment for hypophosphatasia?

While there is currently no cure for hypophosphatasia, a treatment to help manage symptoms is available. In 2015, asfotase alfa (Strensiq™) was approved for use in the US, the European Union, and Canada for pediatric-onset HPP, and in Japan for HPP with onset at any age. The medication is an injection given multiple times per week subcutaneously. It is a recombinant (factory-made) form of ALP that has a bone-targeting component.

For more information, please contact the Soft Bones Foundation
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Finding the Key to HPP