Hypophosphatasia (HPP)

Hypophosphatasia (HPP) is an inherited, progressive, ultra-rare metabolic disease in which patients experience devastating effects on multiple systems of the body and face debilitating or life-threatening complications.[1,2]

Signs and Symptoms Can Include:[1,3-10]

Bones
- Weak or brittle bones
- HPP-related rickets
- Frequent fractures
- Fractures that don't heal properly
- Bowed legs
- Bone pain that won't go away
- Reliance on an assistive device such as crutches, a walker, or a wheelchair

Muscles and Joints
- Muscle weakness
- Arthritis
- Pseudogout caused by deposits of calcium in the joints
- Waddling gait

Ribs and Lungs
- A condition in which bones in the rib cage may not grow or develop properly, leading to underdeveloped lungs, especially in children
- Severe breathing complications that require an assistive breathing device, especially in young children
- Pneumonia

Brain
- A condition in which the bony plate of the skull closes early, causing an abnormally shaped head and potentially causing increased pressure on the brain and swelling of the optic nerve
- Seizures (especially in young children) that can be life-threatening

Kidneys
- A buildup of calcium in the kidneys that may lead to decreased kidney function

Teeth
- Early tooth loss (before the age of five)
- Loss of tooth and its entire root
- Gum disease

HPP is characterized by **defective bone mineralization** that can lead to deformity of bones and other skeletal abnormalities, as well as other systemic complications.[1-3]

New symptoms can appear at any age and symptoms can worsen over time, causing **significant disability**.[1,11]

HPP can affect males and females of all ages and can have **devastating outcomes.**[1,12,13]

Without treatment, **73%** of infants with HPP symptom-onset within the first six months of life **will not survive** beyond five years.[4]

What Causes HPP?

HPP is inherited, resulting from a defect in the gene that makes an enzyme known as tissue non-specific alkaline phosphatase.[1,14] The result is low levels of alkaline phosphatase (ALP) activity.[1,15,16] When ALP functions normally, it allows calcium and phosphate to bind together to form healthy, mineralized bones.[5,14] In patients with HPP, ALP activity is low, leading to improper mineralization.[16] Instead, calcium and phosphate can build up in other places throughout the body, causing damage to bones and organs.[16]

More information about HPP is available at hypophosphatasia.com.

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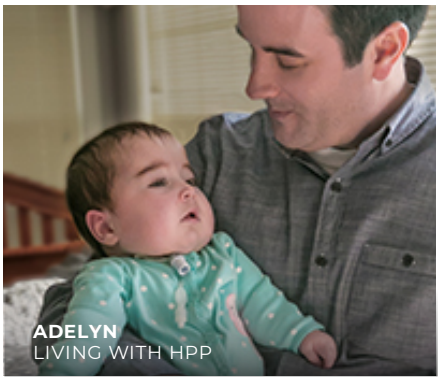
Hypophosphatasia (HPP)

How Is HPP Diagnosed?

- Since HPP shares symptoms with other, more common diseases, diagnostic delays and misdiagnoses are common.[1,17]
- A full clinical assessment and a simple blood test for low ALP can lead to a diagnosis.[1,10]
- It is critical to get an accurate diagnosis as early as possible to ensure that appropriate care is provided.[1,10]

EVIE
LIVING WITH HPP

> **"Evie is our social little butterfly. When we go to the gym, when we go anywhere, everyone knows who Evie is."**
>
> **LINDSEY**
> MOTHER OF EVIE



ADELYN
LIVING WITH HPP

> **"When I hold Adelyn now, it's like holding the sun—a ball of warmth. She just looks at you and warms your heart. I think, while I'm looking at her, that, 'You're amazing. You're a miracle. Here you sit, smiling hugely at me and making my life better each day."**
>
> **GREG**
> FATHER OF ADELYN

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