

# Hypophosphatasia Glossary

## General HPP Terminology

### **Alkaline phosphatase (ALP)**

Enzyme that controls a chemical that blocks skeletal calcification

### **Enzyme**

A protein that breaks down certain chemicals (substrates)

### **Hypophosphatasia (HPP)**

Hypophosphatasia (HPP) is the rare genetic form of rickets or osteomalacia that features paradoxically low serum alkaline phosphatase (ALP) activity.

## Bone Biology

### **Chondrocytes**

Cartilage cells found in growth plates of growing children

### **Collagen**

Most abundant protein in bones, skin, muscles, and tendons

### **Ectopic**

“Out of place or position” (for calcium – outside of the skeleton)

### **Hydroxyapatite**

Crystals of calcium and phosphate deposited into collagen to make bones strong

### **Osteoblasts**

Cells that form bone

### **Osteoclasts**

Cells that break down bone

### **Premature Tooth Loss**

Loss of a “baby” tooth without trauma before the 5th birthday

## Doctors and Medical Specialists involved in HPP Care

### **Endocrinologist**

Diagnoses and treats hormone and metabolic disorders, including HPP

### **Medical Geneticist**

Diagnoses, counsels for, and treats inherited diseases, including HPP

### **Nephrologist**

Treats kidney diseases

### **Neurologist**

Treats brain, seizures, and other nerve problems

### **Ophthalmologist**

Treats eye problems

### **Orthopedist**

Surgeon who manages bone, joint, ligament, muscle, and tendon diseases

### **Physical Therapist**

Provides non-surgical therapy to improve or restore mobility and treat pain

### **Rheumatologist**

Treats inflammation and joint diseases

## Genetics and Genetic Terminology

### **Autosomal Inheritance**

Two most common patterns of disease inheritance

### **Compound Heterozygosity**

Presence of two different copies of a gene

### **Founder Mutation**

Genetic defect from distinct geographical regions

### **Heterozygosity**

One distinctive copy of a gene

### **Genotype**

Genetic make-up of a person

### **Phenotype**

Observable traits in a person as a result of how their genes are expressed

### **Homozygosity**

Presence of identical gene copies

### **Missense (Point) Mutation**

Change of a single DNA

## Possible Complications of HPP and other Bone Disorders

### **Craniosynostosis**

Premature fusion of growth plates in the skull; can cause pressure on the brain and require surgery

### **Hypercalcemia**

Elevated blood calcium

### **Microfractures**

Microscopic fractures

### **Nephrocalcinosis**

Calcium-phosphate (mineral) in the kidneys

### **Rickets**

Softening and weakening of bones during growth from too little hydroxyapatite deposition

### **Osteoarthropathy**

Loss of joint cartilage causing arthritis

### **Osteomalacia**

Softening of bones from too little deposited hydroxyapatite crystals

### **Pseudofractures**

X-ray feature of osteomalacia (soft bones) in adults

### **Rachitic Chest**

Pulled-in sternum or “pigeon breast” deformity from rickets (soft bones during growth)

## **Tests**

### **Alkaline Phosphatase Blood Test**

Measures ALP activity in blood serum. Patients with HPP have low ALP levels for their age

### **ALPL Gene**

Mutation (defect[s]), 1 or 2, in this gene underlie HPP

### **Bone Biopsy**

Taking a small sample for microscope examination

### **Bone Scan**

Small amount of radioactive dye injected into a vein that detects where abnormalities are in the skeleton

### **Chemicals (Substrates) That Accumulate in HPP**

- Pyridoxal 5' phosphate (PLP): a form of Vitamin B6
- Inorganic pyrophosphate (PPi): the blocker of bone mineralization
- Phosphoethanolamine (PEA): often elevated in HPP blood or urine

### **Osteomalacia**

Softening of bones from too little deposited hydroxyapatite crystals

### **DXA (DEXA)**

X-ray to measure bone mineral density

### **PTH and 25-Hydroxyvitamin D**

Hormones that regulate blood calcium levels

### **Radiograph**

An x-ray

### **Osteomalacia**

Softening of bones from too little deposited hydroxyapatite crystals

### **Renal (Kidney) Ultrasound**

Sonar method to check for kidney calcification or stones

### **Serum Calcium**

Blood test to diagnose or monitor blood calcium level for many bone diseases

## Treatment of HPP

### **Asfotase Alfa (trade name Strensiq™)**

ALP replacement therapy given by injection. Approved to treat pediatric-onset HPP

### **Supportive**

Good dental care, analgesics if sufficient for mild occasional aches or pains

## Types of HPP

### **Adult Hypophosphatasia**

Typically presents in middle age; sometimes misdiagnosed as osteoporosis

### **Benign Prenatal Hypophosphatasia**

Common pediatric HPP. Skeletal changes (limb shortening and bowing) in the womb or noted at birth, and then improving spontaneously. Not as severe as perinatal HPP

### **Childhood Hypophosphatasia**

Manifests after age 6 months and before age 18 years

### **Hypophosphatasia Carrier**

Has single HPP mutation, but is healthy

### **Infantile Hypophosphatasia**

Manifests after birth but before age six months

### **Odontohypophosphatasia**

Mildest HPP: Tooth problems only

### **Perinatal Hypophosphatasia**

Most severe HPP; strikingly apparent at birth

### **Pseudohypophosphatasia**

Extremely rare. Like infantile HPP, but blood ALP normal or elevated

For more information, please contact the Soft Bones Foundation

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