

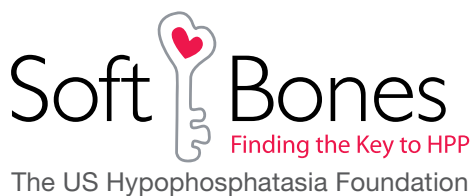
## WHAT IS HYPOPHOSPHATASIA

Hypophosphatasia (HPP) is a metabolic bone condition that most often affects the development of bones and teeth. The signs and symptoms vary widely, with mild cases causing only dental abnormalities while the more severe types may have life-threatening consequences.

The clinical hallmark of the disorder is premature tooth loss and bones that become soft or weakened, sometimes causing skeletal deformities and fractures.

Because the severity of HPP varies greatly, it is important for the patient, family, caregivers, and health professionals to understand the individual patient's level of disease to provide appropriate care.

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## Physical and Occupational Therapy Interventions for Patients with Hypophosphatasia

### Role Of Physical And Occupational Therapy

Physical Therapy (PT) and Occupational Therapy (OT) can serve important roles in the evaluation, education, treatment, and management of HPP. The goal of PT/OT intervention is to maximize function, including mobility and independence. Individualized guidelines can be established for care in the home, school, and community settings.

### Evaluation

Once HPP has been diagnosed by the physician, a referral to PT/OT may be recommended to evaluate movement and developmental progression in infants and children. In HPP adolescents and adults, PT/OT evaluations focus on joint mobility, activities of daily living (ADLs), and endurance with prolonged sitting or walking. Standardized assessment tools are important in determining if further intervention by a therapist is needed.

### Education

The role of the therapist begins with educating the patient and family concerning how HPP can compromise movement, endurance, alignment, and mobility. Findings from the evaluation are discussed and provide a baseline or “starting point” for therapy. In the event that problems are identified, treatment goals are established, and further interventions may be recommended.

### Treatment

Treatment interventions are individualized based on the clinical findings and the age/stage of development of the patient. Please see the tables on pages 2 and 3 for these recommended treatment interventions.



## TREATMENT INTERVENTIONS INFANTS AND CHILDREN

Clinical findings that may be present in infants and children with HPP	Therapeutic Interventions
Bony Fragility	<ul style="list-style-type: none"> <li>• Education to caregivers in safe handling, positioning, and transferring to minimize the risk of fracture during feeding, bathing, dressing, and diaper changes</li> </ul>
Pain	<ul style="list-style-type: none"> <li>• Education to caregivers in observing signs of pain in the infant which may indicate the need for modification of position or activity</li> </ul>
Respiratory Compromise	<ul style="list-style-type: none"> <li>• Under direction of physician, a therapist may position to provide postural drainage and chest physical therapy via percussion or vibration</li> </ul>
Gross Motor Delay	<ul style="list-style-type: none"> <li>• Provide safe and developmentally appropriate positioning to encourage strengthening of arms, legs, head, and trunk</li> <li>• Provide opportunity for visual interaction and manual manipulation of toys</li> <li>• Ensure supportive seating, orthotics, and standing devices under direction of physician to progress to upright and weight bearing activities</li> <li>• Educate family and caregivers in daily home program</li> </ul>
Low Bone Density/ Delayed Mobility	<ul style="list-style-type: none"> <li>• Assess need for:               <ul style="list-style-type: none"> <li>– orthotics (braces, shoe inserts)</li> <li>– assistive device (walker, crutches)</li> <li>– wheelchair</li> </ul> </li> <li>• Develop weight bearing program to improve bone health</li> <li>• Progression to walking, as able</li> <li>• Provide guidelines and limitations on gross motor activity to prevent injury in the presence of bony fragility</li> </ul>
Presence of Skeletal Deformity	<ul style="list-style-type: none"> <li>• Positioning to prevent deformity and maintain joint motion</li> <li>• Splinting and orthotics to provide support, positioning, and joint protection</li> </ul>
Muscle Weakness	<ul style="list-style-type: none"> <li>• Provide safe and developmentally appropriate positioning to encourage strengthening of arms, legs, head, and trunk</li> <li>• Provide exercise and activities for strengthening</li> <li>• Develop low impact home exercise program and safe extracurricular activities</li> <li>• Modification of ADLs and handwriting tools may assist if weak grip and pinch present</li> </ul>
Low Endurance /Activity Limitations	<ul style="list-style-type: none"> <li>• Minimize unnecessary distances and stairs</li> <li>• Modification of the environment to keep items close that are frequently used</li> <li>• Ensure school setting modified with second set of books to prevent carry; modify physical endurance activities as needed</li> <li>• Use of walker, crutches or wheelchair, as needed</li> <li>• Provide resources for modified sports participation</li> </ul>

## TREATMENT INTERVENTIONS ADOLESCENTS AND ADULTS

Clinical findings which may be present in Adolescents & Adults with HPP	Therapeutic Interventions
Joint Limitation	<ul style="list-style-type: none"> <li>• Educate on specific bone health and joint limitations</li> <li>• Modify tasks and positioning to accommodate joint limitation; especially with prolonged sitting</li> </ul>
Bone Pain/Limited Endurance	<ul style="list-style-type: none"> <li>• Functional and strengthening exercises to support bone</li> <li>• Educate on low impact activity</li> <li>• Minimize stress to joints and spine</li> <li>• Utilize orthotics, and/or assistive device as needed to decrease pain and provide support and balance for ambulation</li> <li>• Wheelchair for extended distances, as needed</li> <li>• Modification of environment to ensure efficiency of movement for task completion</li> </ul>
Limitations with ADLs	<ul style="list-style-type: none"> <li>• Improve grip and pinch strength through exercise</li> <li>• Evaluate need for ADL tools: reacher, dressing aides, pencil and pen grips; bottle jar openers; grab bar and railings and tub chairs</li> </ul>
Obesity	<ul style="list-style-type: none"> <li>• Provide education for healthy weight to protect the skeleton and joints</li> <li>• Set goals for safe aerobic activity</li> <li>• Educate for healthy living and caloric intake</li> </ul>

### Monitoring and Reassessment

Follow-up may be recommended more frequently for infants and growing children to ensure developmental progression. For an adult or adolescent, re-evaluation may be recommended by the physician depending on how the disease affects function.

### Pharmaceutical Treatment

In 2015, asfotase alfa (Strensiq™) became the only approved medical treatment for use in the US, the European Union, and Canada for pediatric-onset HPP, and in Japan for HPP with onset at any age. This therapy has greatly improved the function of HPP patients who are appropriate candidates for this treatment.

### How can therapy be helpful in patient's receiving pharmaceutical treatment?

The role of the physical and occupational therapist continues to be important for many patients with HPP. Standardized testing to measure strength, endurance, function, and quality of life, both before and after initiation of medical treatment, helps to assess patient progress.