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Opinion Article

Clinical Features and Management Strategies of Hypophosphatasia

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ABOUT THE STUDY

Hypophosphatasia (HPP) is a rare inherited metabolic disorder characterized by deficient activity of the Tissue-Nonspecific Alkaline Phosphatase (TNSALP) enzyme. This enzyme plays a crucial role in the metabolism of inorganic Pyrophosphate (PPi) and Pyridoxal 5'-Phosphate (PLP), which are essential for bone mineralization and several other biological processes. The deficiency of TNSALP leads to the accumulation of PPi and PLP, resulting in abnormal skeletal development and mineralization. The clinical presentation of HPP can vary widely, ranging from mild dental abnormalities to severe skeletal deformities and life-threatening complications.

Clinical features

The clinical manifestations of HPP can be categorized into six major forms based on the age of onset and severity: perinatal lethal, prenatal benign, infantile, childhood, adult, and odontohypophosphatasia. Here are the clinical features associated with each form:

Perinatal lethal HPP: This is the most severe form and is characterized by marked hypomineralization of the skeleton, respiratory distress, and usually results in stillbirth or early neonatal death.

Prenatal benign HPP: This form is typically associated with mild skeletal abnormalities, such as bowing of the long bones, and is usually identified incidentally on prenatal ultrasound. The prognosis is generally good, and affected individuals often show spontaneous improvement after birth.

Infantile HPP: This form presents within the first six months of life with failure to thrive, respiratory difficulties, rickets, and often progressive skeletal deformities, such as craniosynostosis, short stature, and enlarged joints.

Childhood HPP: Children with this form typically present with premature loss of deciduous teeth, skeletal abnormalities, and growth delays. Fractures, dental abscesses, and bone pain are common features.

Adult HPP: This form may present at any age and is characterized by musculoskeletal pain, stress fractures, and dental problems. Individuals with adult HPP may experience recurrent fractures and chronic pain that significantly impact their quality of life.

Odontohypophosphatasia: This form primarily affects the teeth, leading to dental abnormalities, such as premature loss of primary and permanent teeth, enamel hypoplasia, and spontaneous dental abscesses.

Management strategies: The management of HPP requires a multidisciplinary approach involving specialists in genetics, endocrinology, orthopedics, dentistry, and supportive care. The management strategies for HPP primarily focus on symptom control and optimizing quality of life. Here are some key aspects of HPP management:

Symptomatic treatment

Pain management: Nonsteroidal Anti-Inflammatory Drugs (NSAIDs) or opioids may be prescribed to manage musculoskeletal pain.

Fracture prevention: Measures to minimize the risk of fractures, such as fall prevention strategies, physical therapy, and assistive devices, should be implemented.

Dental care: Regular dental evaluations and appropriate interventions, such as fluoride treatments, oral hygiene measures, and dental restorations, can help maintain oral health.

Nutritional support

Adequate calcium and vitamin D supplementation: These nutrients are essential for skeletal health and may be prescribed to support bone mineralization.

Nutritional assessment and support: Monitoring nutritional status, especially in infants and children, and providing appropriate nutritional interventions can optimize growth and development.

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Enzyme Replacement Therapy (ERT)

Asfotase alfa: This recombinant human TNSALP enzyme replacement therapy has been approved for the treatment of perinatal and infantile-onset HPP. It helps normalize the disturbed mineral metabolism and improve skeletal mineralization. Regular intravenous injections are required.

Surgical interventions

Orthopedic procedures: Surgical interventions may be necessary to address skeletal deformities, correct craniosynostosis, or manage fractures that do not respond to conservative measures.

Dental interventions: Prosthetic dental solutions, such as dentures or dental implants, may be required for individuals with severe dental abnormalities.

Genetic counseling

Genetic counseling should be provided to affected individuals and their families to discuss the inheritance pattern of HPP, recurrence risks, and the availability of prenatal testing for future pregnancies.

Hypophosphatasia is a rare inherited metabolic disorder characterized by deficient tissue-nonspecific alkaline phosphatase enzyme activity, leading to impaired skeletal mineralization.

The clinical features of HPP vary widely, and management strategies aim to alleviate symptoms, prevent complications, and optimize quality of life. Early diagnosis, a multidisciplinary approach, and targeted interventions can contribute to better outcomes for individuals with HPP.