Hypophosphatasia: Dental Manifestations and Guiding Treatment Planning

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Abstract

Hypophosphatasia (**HPP**) is an uncommon inherited disorder which can lead to growth and development defects ranging from stillbirth to early exfoliation of primary dentition. Different forms of hypophosphatasia have differing inheritance patterns and degrees of symptom severity. Overarchingly, the condition causes defective bone and tooth mineralization. Mutations in the ALP gene for such patients lead to low activity of the tissue nonspecific alkaline phosphatase enzyme.

This presentation follows a child who has been an active patient at the Rutgers School of Dental Medicine's pediatric clinic for over 2 years. The patient had been diagnosed with hypophosphatasia at a young age and has a robust interdisciplinary healthcare team, including a pediatric dentist. This case report will detail the clinical and radiographic findings of hypophosphatasia as well as treatment planning modifications that are indicated in the highlighted demographic.

Background

Disease Etiology

- Rare genetic metabolic disease
- Impaired mineralization
- ALP enzyme is defective (important for bone development)
- Ca and PO4 build up elsewhere in the body → damaging organs → weakness and deformity in bones & premature loss of teeth

Manifestations

- **Systemic**
  - Failure to thrive
  - Brittle bones and lack of muscular development
  - Chest deformities
  - Waddling gait
  - Frequent bone fractures
  - Pain in Achilles tendon, rotator cuff and elbow
- **Intraoral**
  - Premature unexplained primary dentition exfoliation (Irregular cementum)
  - Irregular/Sharp tooth crown anatomy
  - Bone loss and generalized increase in mobility

Disease Management

Early Diagnosis

From a pediatric dentist's perspective premature loss of primary dentition before the age of 5 years old (not due to extensive caries or trauma) can signal to a provider that patient may have systemic pathology linked to such oral manifestations. Radiographic findings as well as a history of fractures have often been accompanied with HPP.

Further workout with blood tests and genetic assays can help narrow the differential to HPP.

Medical Intervention

Medications include use of growth hormone treatment. Strensiq (asfotase alfa) is an enzyme replacement therapy for patients with perinatal/infantile onset HPP.

Dental Management

- Consider more frequent recall appointments
- Detailed history of trauma and etiology of any premature primary dentition loss
- Caution exercised during exam and prophies (aggressive scaling can cause further loss of teeth that present with early bone loss and mobility)
- Select case reports slow excessive bone loss while others detail HPP-medication induced ankylosis
- Consider appliances to tooth replacement (dependent on existing dentition's periodontal support)
- Increased risk of periodontal disease

Normal Phosphate Levels in the Human Body
What functions are carried out by PO4?

- DNA synthesis
- Energy metabolism
- Bone and teeth mineralization

Seek a pediatric dental professional:

- Age 1 year old or 6 months after eruption of first primary tooth
- Premature loss of primary dention (no associated trauma)
- Medically confirmed diagnosis of HPP

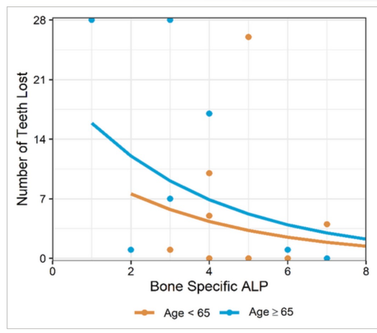
Conclusion

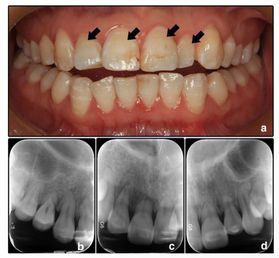
Taking a thorough history (assessment of trauma, fractures, muscle weakness alongside dental examination and radiographic evaluation) is key in early detection. Patients with confirmed or suspected HPP should be placed on more frequent dental recalls. Caution should be taken when treating patients to avoid use of excessively pressure (ex. While scaling) which can cause further mobility and loss of teeth.

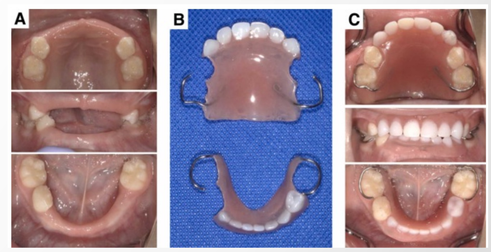
Diligent dental examinations with proper history/workup can help prevent such missed diagnoses, allowing patients and their caregivers to seek medical attention as early as possible.

Future Direction

Further research on the early diagnostic markers is necessary. Many times, premature loss of primary dentition is incorrectly equated to traumatic avulsion and medical blood test references are calibrated to adult serum levels, therefore missing characteristically low phosphate levels in children.







Acknowledgments

Sincere gratitude for Dr. Mohan, our clinical director and case report mentor. Special thank you to our patients and clinical staff at Rutgers School of Dental Medicine.