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Case Report

Dental and oral manifestations in a patient with hypophosphatasia: a case report

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ABSTRACT

Hypophosphatasia (HPP) is an uncommon inherited metabolic disorder caused by deficient activity of tissue-nonspecific alkaline phosphatase (TNAP), resulting in impaired mineralization of skeletal and dental tissues. Dental manifestations are often among the earliest clinical signs and may include premature tooth loss, enamel and dentin defects, and delayed eruption. A 13-year-old female with a confirmed diagnosis of infantile-onset congenital HPP presented with pain in the upper left posterior region. She was receiving enzyme replacement therapy with asfotase alfa along with growth hormone supplementation. Clinical and radiographic examination revealed a non-vital maxillary first molar with a defective restoration, generalized enamel and dentin defects, periodontal involvement, mobility of anterior teeth, delayed and ectopic eruption, and malocclusion characterized by an anterior open bite with midline deviation. Endodontic management followed by definitive restoration of the affected tooth successfully resolved the patient's symptoms. A comprehensive dental care plan including oral hygiene reinforcement, caries control, and extraction of non-restorable teeth was instituted, with long-term follow-up planned. This report highlights the importance of early diagnosis and coordinated multidisciplinary dental care in patients with HPP.

Keywords: Hypophosphatasia, Enamel hypoplasia, Premature tooth loss, Asfotase alfa, Pediatric dentistry

INTRODUCTION

Hypophosphatasia (HPP) is a rare inherited metabolic disorder resulting from mutations in the ALPL gene, which encodes TNAP. TNAP plays a critical role in the mineralization of bone and teeth by regulating inorganic pyrophosphate levels, a natural inhibitor of hydroxyapatite formation.[1] Deficiency of this enzyme leads to defective mineralization of hard tissues and a wide spectrum of clinical manifestations.[2] The clinical presentation of HPP ranges from severe perinatal forms to milder variants that predominantly affect dental tissues, known as odonto-HPP.[3] Skeletal involvement may include rickets, osteomalacia, fractures, and growth disturbances, whereas dental manifestations often present earlier and may occur in the absence of overt skeletal signs.[4] Premature exfoliation of fully rooted primary teeth due to defective acellular cementum formation and impaired periodontal attachment is considered a hallmark dental feature of HPP. Additional oral findings include enamel hypoplasia, thin dentin, enlarged pulp chambers, root abnormalities, periodontal disease, delayed or ectopic eruption, tooth mobility, and malocclusion.[5] These features frequently necessitate long-term multidisciplinary dental management.

Recent advances in enzyme replacement therapy, particularly the introduction of asfotase alfa, have significantly improved survival and skeletal outcomes in patients with severe forms of HPP. However, dental

manifestations often persist, especially when mineralization defects develop before initiation of therapy.[6] This case report describes the dental and oral features of hypophosphatasia in an adolescent patient and discusses management considerations in light of current evidence.

CASE REPORT

A 13-year-old female presented to the pediatric dental clinic with her parents, reporting pain in the upper left posterior region. Her medical history was significant for infantile-onset congenital hypophosphatasia. She was undergoing enzyme replacement therapy with asfotase alfa (68 mg administered subcutaneously three times weekly) and was also receiving growth hormone therapy.

Intraoral examination revealed a large defective restoration in the maxillary left first molar tooth 26 (Figure1) which was tender on percussion and diagnosed as non-vital. Additional findings included generalized enamel and dentin defects (Figures 2 and 3), periodontal involvement, increased mobility of anterior teeth, delayed and ectopic eruption of permanent teeth, and malocclusion characterized by an anterior open bite with right-sided midline deviation. Radiographic examination confirmed these findings.

Root canal treatment followed by definitive coronal restoration of tooth 26 was performed, resulting in complete resolution of the patient's symptoms. A comprehensive dental management plan was formulated, including reinforcement of oral hygiene measures, caries control strategies, and extraction of non-restorable teeth. The patient was scheduled for regular follow-up visits to monitor oral health and disease progression.

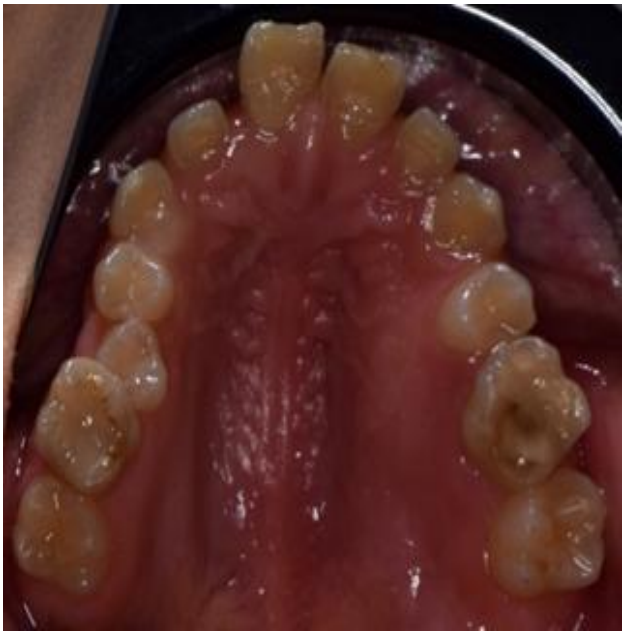

Figure 1: Clinical photograph showing defective restoration in tooth #26.

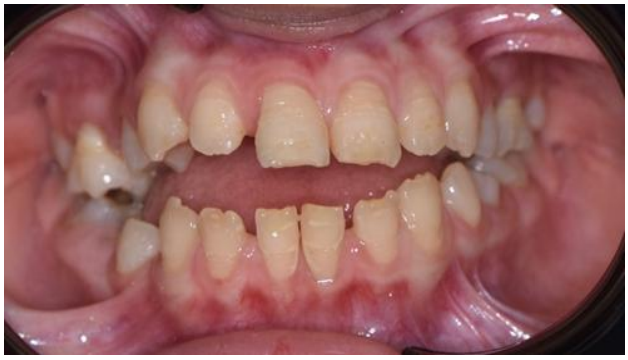

Figure 2: Frontal clinical photograph showing enamel hypoplasia in all upper and lower teeth.

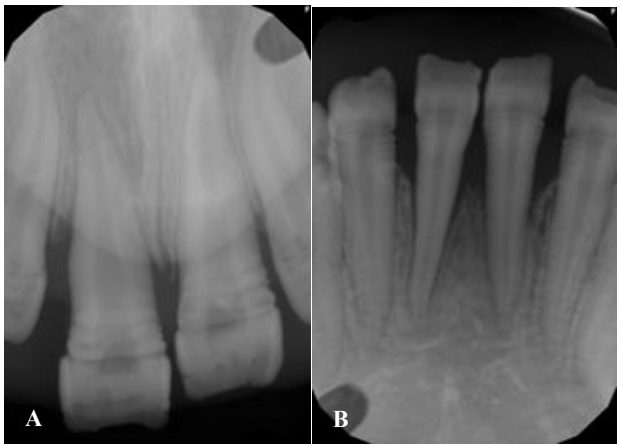

Figure 3 (A and B): Radiographic image showing anomalies in the upper and lower anterior teeth.

DISCUSSION

Dental manifestations represent a prominent and often early feature of hypophosphatasia. Enamel and dentin defects, periodontal disease, abnormal tooth mobility, and eruption disturbances are well-documented findings. The clinical features observed in the present case align with previously reported phenotypes in pediatric HPP populations.[6] The combination of enamel and dentin hypomineralization with compromised periodontal support increases susceptibility to dental caries, pulp pathology, and premature tooth instability. In the present case, the necrosis of the maxillary first molar underscores the heightened risk of endodontic complications associated with structurally compromised dental tissues.[3,4] Malocclusion, delayed eruption, and midline deviation observed in this patient are consistent with reports describing altered craniofacial growth and dentoalveolar development in HPP.[2] Defective alveolar bone remodeling further contributes to occlusal disturbances and challenges orthodontic management.[7]

Although enzyme replacement therapy with asfotase alfa has demonstrated sustained improvements in skeletal mineralization and overall function, its effect on dental tissues remains variable.[8] Dental defects established prior

to treatment initiation are largely irreversible, necessitating ongoing preventive and restorative dental care. The concurrent use of growth hormone therapy in this patient may have contributed to overall somatic growth.[9,10] However, its direct influence on dental mineralization remains unclear and warrants further investigation.

Radiographic findings such as reduced alveolar bone height, delayed eruption, and root abnormalities are characteristic of hypophosphatasia and correlate with clinical instability of teeth.[11] Early identification of these features allows timely intervention and long-term planning. Multidisciplinary collaboration remains essential to preserve oral function and quality of life in affected individuals.

CONCLUSION

This case illustrates the complexity of dental management in patients with hypophosphatasia, particularly those with infantile-onset disease who continue to exhibit oral manifestations into adolescence. The patient presented with multiple dental challenges including pulp necrosis, enamel and dentin defects, periodontal involvement, and malocclusion, reflecting the systemic impact of impaired mineralization. Successful endodontic intervention addressed the immediate complaint, while a comprehensive long-term care plan was established to manage ongoing dental needs. Early recognition of oral manifestations and close integration of dental and medical care are critical to optimizing outcomes in patients with hypophosphatasia.

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