Prosthetic rehabilitation of a young patient with Hypophosphatasia - A review and case report

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Abstract

Hypophosphatasia is a congenital disease characterized by deficiency of serum and tissue non-specific alkaline phosphatase activity. The disease occurs due to mutations in the liver/bone/kidney alkaline phosphatase gene. Six clinical forms of hypophosphatasia are recognized. Systemic symptoms of the disease are respiratory complications, premature craniosynostosis, widespread demineralization and rachitic changes in the metaphases, stress fractures, chondrocalcinosis and osteoarthropathy. Characteristic dental symptoms include premature loss of deciduous teeth, premature exfoliation of fully rooted permanent teeth, severe dental caries and alveolar bone loss. This clinical report describes the prosthetic rehabilitation of a sixteen year old female patient with hypophosphatasia with partial anodontia and no occlusion. Patient was managed clinically by saving her remaining teeth, fabricating a maxillary overdenture and mandibular cast partial denture. This not only helped her functionally in biting and chewing food and achieving a good occlusion, but also restored her confidence and self image by improving the aesthetics.

Keywords: Alkaline phosphatase levels, Alveolar bone atrophy, anodontia, complete rehabilitation, hypophosphatasia, serum

Introduction

Hypophosphatasia is a rare enzymopathy that normally presents within the first few years of life and often has profound effects upon the periodontium. It is characterized by low or zero levels of serum and tissue non-specific alkaline phosphatase, which is necessary for normal bone mineralization. It is a heritable disorder characterised by defective mineralisation of the skeletal and dental structures of the body, and a deficiency in liver/bone/kidney isoenzyme of alkaline phosphatase (ALP). The symptoms are highly variable in their clinical expression, ranging from stillbirth without mineralized bone, to early loss of tooth without any bone symptoms.[1] In 1957, Fraser[2] classified hypophosphatasia into 3 groups depending upon age of presentation. Six clinical types of hypophosphatasia are currently recognized in the forms of perinatal lethal, benign prenatal, infantile, childhood, adult and odontohypophosphatasia. Some infants can only live for a few days and have respiratory complications due to hypoplastic lungs and rachitic deformities of the chest.

Dental symptoms/Oral manifestations

A few oral manifestations of hypophosphatasia can be given as follows:

- Lack of cementum formation
- Root resorption
- Enamel hypoplasia
- Delayed appearance and premature decay of deciduous and permanent dentition and total tooth loss before 20 years of age.

There appears to be no male/female preference, and mean age for the first tooth to be lost is 21-22 months.[3] Enlarged pulp spaces and reduced cortical bone thickness of the mandibular angle are one of the common characteristics. Roots either fail to develop fully or there is early resorption of the apices.[4]

Radiographic features

Long bones with elongated finger like processes of uncalcified tissue extending longitudinally into the poorly formed osseous tissue at the ends of long bones can be seen radiographically. “Beaten copper” appearance – multiple radiolucent areas on skull called gyral or convoluted markings which result due to increase in intra cranial pressure are seen. Reduced enamel thickness, enlarged pulp chamber and alveolar bone atrophy are common dental findings along with generalised reduction in bone density in jaw bones. Evidence of skeletal defects is found in nearly all patients and includes hypomineralization, rachitic changes, and incomplete vertebrate ossification and occasionally lateral bony spurs on the ulnae and fibulae.[5]

Laboratory diagnosis

The pathognomonic symptom is subnormal serum activity...
of ALP. In general, the clinical severity mirrors the degree of enzyme deficiency. The most sensitive substrate marker for hypophosphatasia is an increased pyridoxal 5'-phosphate (PLP) plasma level, which often correlates with the disease severity. And, although it remains only a research technique, quantitation of urinary inorganic pyrophosphate (PiP) levels, which are elevated in most hypophosphatasia patients, has been reported to accurately detect carriers. In addition, increased urinary levels of Phosphoethanolamine (PEA) are observed in most patients. Differential diagnosis for this disease include Osteogenesis imperfect, Rickets and Achondrogenesis.\(^{[5,6]}\)

**Case report presentation**

This clinical report describes the prosthetic rehabilitation of a sixteen year old female patient with hypophosphatasia, who reported to our department with poor oral hygiene, missing teeth, unaesthetic appearance and low self esteem. Clinical examination revealed severely resorbed maxillary and mandibular alveolar ridges with partial anodontia and several root stumps. Teeth present were right maxillary central incisor, maxillary right first and second molar, maxillary left first and second molar, mandibular left first and second molar, and mandibular right first and second molar. Patient did not have any established occlusion with poor aesthetics [Figures 1-2]. Orthopantomogram (OPG) and a Lateral Cephalogram were taken. OPG revealed severe alveolar bone loss with enlarged pulp chambers and stunted root growth of molars. There were horizontally impacted third molars and reduced bone density with severe bone atrophy in both maxilla and mandible. [Figure 3] On examining the Lateral Cephalogram, a prognathic relation of mandible with maxilla was observed [Figure 4].

**Laboratory testing**

The patient was tested for ALP levels, which were found to be 3 times lower than the normal values confirming the diagnosis of hypophosphatasia. Long bone radiographs appeared normal. Treatment for this patient was planned keeping in mind her psychological aspect, young age and severe bone loss with partial anodontia. It was carried out step by step in various phases: rehabilitating the periodontal condition with non-surgical intervention that included complete oral prophylaxis [Figures 1-2]. Orthopantomogram (OPG) and a Lateral Cephalogram were taken. OPG revealed severe alveolar bone loss with enlarged pulp chambers and stunted root growth of molars. There were horizontally impacted third molars and reduced bone density with severe bone atrophy in both maxilla and mandible. [Figure 3] On examining the Lateral Cephalogram, a prognathic relation of mandible with maxilla was observed [Figure 4].
and curettage; extractions of the unwanted root stumps; and, root canal treatment of 11, 16 and 27. Patient was thus prepared for prosthetic intervention to replace missing teeth, improve the jaw relation and establish a comfortable working occlusion for the young girl. Besides, her extraoral features like sunken cheeks, depressed mentolabial sulcus and decreased nasolabial angle were also kept in mind while fabricating the dentures. As the alveolar ridges were severely resorbed, conventional complete denture or partial denture fabrication was ruled out. Implant retained prosthesis was also not an option as bone height was very less due to resportion, and the distance between the bone crest and the occlusal level was more, which would have led to an unfavourable crown: implant ratio. Hence, it was decided to restore the missing teeth and occlusion with maxillary overdenture and mandibular cast partial denture, as this was the most favourable and conservative option for this young girl with severe bone loss and lost psychological confidence.

After the pre-treatment procedures, the tentative aesthetic anterior try-in was performed [Figure 5] and improved aesthetics with adequate lip fullness and improved inter arch relationship was observed. The root canal treated teeth was carried out for the abutment teeth (11, 16 and 27), followed by the fabrication of post integrated metal copings [Figure 6]. Mandibular cast partial metal framework was planned with a ring clasp placed on 37 and embrasure clasps on 46 and 47. Mesh framework was designed to cover the edentulous ridge span [Figure 7]. Following this, jaw relation and final try-in was done. The regular procedures for overdenture and cast partial denture fabrication were carried out in the lab, followed by final denture insertion [Figure 8]. After placement of the dentures, we could appreciate the restored aesthetics, improved occlusion and an established vertical dimension for this young girl with a better functional occlusion that improved her speech, mastication and most importantly, her self confidence. The post operative frontal and lateral profile view depicted an improvement over the depressed mentolabial sulcus, and an increased and more acceptable nasolabial angle and cheek bulging, with an improved normal interjaw relationship. [Figures 9 - 10]  Patient was more confident with her improved aesthetic smile and restored occlusion.
Outcome of the treatment. Thus, to conclude, a patient with hypophosphatasia poses a challenge for the clinician, especially when the patient is of young age as such patients have high expectations; however, a thorough diagnosis, constant motivation and critical evaluation of all treatment options and correct treatment planning and selection can lead to a successful treatment for our patients. This case presentation is different as it presents a complete intra as well as extra oral rehabilitation of a young girl with hypophosphatasia, and with severely resorbed ridges, in whom conventional treatment planning was not possible. Besides, managing the aesthetics, function and occlusion along with improving the extra-oral appearance of the patient posed a great challenge.

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