Oral and Dental Findings in Osteopetrorickets

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While dental findings of both rickets and osteopetrosis have been reported, there is no published report on the oral and dental findings of osteopetrorickets. In this paper dental findings of osteopetrorickets were presented. A two-year-old female child was referred to the pedodontics clinic for dental examinations before bone marrow transplantation. Her teeth showed severe mobility and the eruption of the teeth were delayed. The dental findings of the patient were different from that of osteopetrosis and rickets.
Key words: *Osteopetrosis, Rickets, Osteopetrorickets, Oral and dental findings*
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INTRODUCTION

Osteopetrosis is a rare skeletal condition. Its characteristic finding is skeletal sclerosis caused by aberrant osteoclast-mediated bone resorption. Three clinically distinct forms of osteopetrosis are recognized; the infantile malignant autosomal recessive form, the intermediate autosomal recessive form, and the adult benign autosomal dominant form. The disease represents a spectrum of clinical variants because of heterogeneity of genetic defects resulting in osteoclast dysfunction.[1]

The major clinical features of infantile malignant form of the disease derive from failure of resorption of bone and consequently abnormally dense of bone. Progressive encroachment on the major foramina of the skull leads to compression of optic and acoustic cranial nerves and causes to blindness and/or deafness. Bone marrow transplantation with appropriately human leukocyte antigen (HLA)-matched donor is the only possible treatment option for malignant osteopetrosis.[2,3]

Rickets is characterized by undermineralized bone matrix and results from abnormalities of vitamin D and deficiency of calcium and phosphorus. It is an X-linked dominant disease and characterized by a renal defect leading to reduced phosphate resorbtion in the glomerular filtration system of proximal renal tubules. This deficiency leads to hypophosphatemia and hyperphosphaturia that produces bone and dental abnormalities.[4] In some patients with osteopetrosis, paradoxical rickets have been reported. It probably results from inability of the osteoclasts to maintain a normal calcium phosphorus balance in the extracellular fluid, despite positive total body calcium balance.[2,5] The term "Osteopetrorickets" is first used in 1995 by Kaplan et al. to define this unusual association.[5]

Dental and intraoral findings of both osteopetrosis and rickets have been reported in detail. Oro-dental findings of osteopetrosis include delayed eruption, congenitally absent teeth, unerupted and malformed teeth, and enamel hypoplasia. Decreased alveolar bone production, defective and abnormally thickened periodontal ligament have also been reported.[6-14] Osteomyelitis is a serious complication of this disease, and occurs most frequently in mandible and occasionally in maxilla, scapula, and extremities.[11,12] In addition to these clinical reports in humans, a plethora of animal studies on osteopetrosis have also been published. In these studies, similar findings have been reported in osteopetrotic mice and rabbits.[9,10,13]

Dental findings of rickets include hypoplastic enamel and dentin, chronic periodontal disease, and multiple dental abscesses. In this disease, periapical radiolucencies, abscess, and fistulas associated with micro exposures can be seen in either the primary or the permanent teeth. Rickets is especially characterized by pulpal horns extending to dentinoenamel junction. Late eruption of teeth is also reported. Apical closure is delayed, especially in permanent teeth.[4,6-8,14]

Although clinical, biochemical, and radiographical findings have been well described, intraoral and dental findings of osteopetrorickets have not been reported previously. This report presents a case of osteopetrorickets in a 2-year-old girl and describes the oral and dental findings of the disease.

CASE REPORT

A 2-year-old girl was referred to the Pedodontics clinic. She had been followed the department of Pediatrics and was sched-

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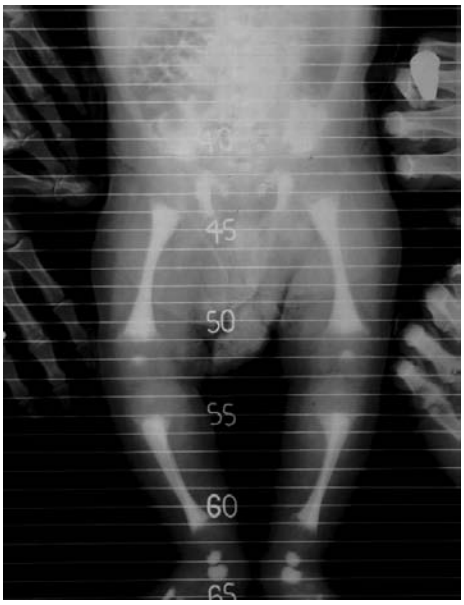

Figure 1: Skeletal radiograph of the patient

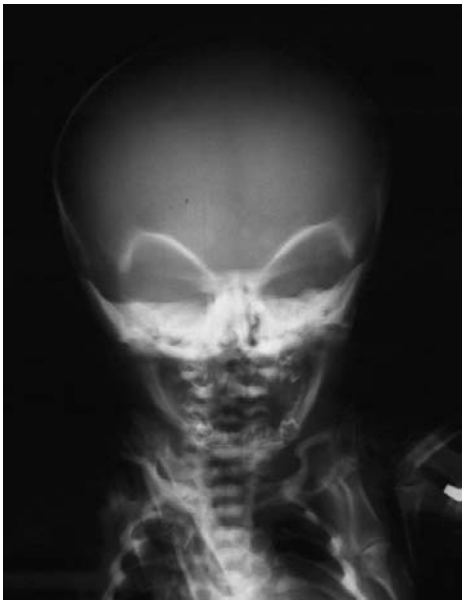

Figure 2: Thickening of the base of the skull and "mephistophelean" appearance of the orbits

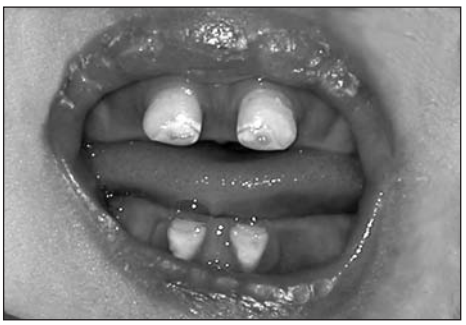

Figure 3: Oral and facial (small picture) appearance of the patient

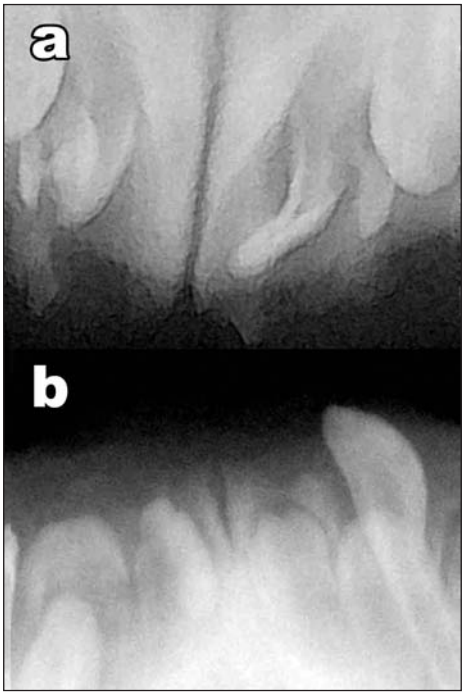

Figure 4: Radiographs of upper and lower incisors

uled for bone marrow transplantation. Before commencement of the transplantation, the parents were asked to complete any possible dental treatment. The patients were in excellent health and did not have a history of consanguineous mating. The patient was diagnosed for rickets at her first referral to the hospital when she was 3-months-old.

She had frontal bossing, craniotabes, rachitic rosary, hepathosplenomegaly, hypotonia (lack of head control), and nystagmus. The further radiographic examinations revealed diffuse bonny sclerosis with defective methaphyseal modeling and a "bone in bone" appearance, loss of demarcation of the cortex and medullary cavities in skeletal radiographs (Figure 1). Thickening of the base of the skull and "mephistophelean" appearance of orbits, which were typical for osteopetrosis, were observed in her craniofacial radiographs (Figure 2). She was diagnosed with osteopetrorickets following clinical, biochemical and radiographic examinations; after which bone morrow transplantation was indicated.

Intraoral examination showed that teeth 51, 61, 71, 81 had erupted. However, they were severely mobile. The enamel of teeth was hypoplastic and presented with a yellowish shade. Eruption of the teeth was delayed (Figure 3). Periapical radiographs showed unerupted and abnormally shaped teeth (Figure 4). The teeth 51, 61, 71, 81 were extracted due to severe mobility as well as to prevent the patient from a possible aspiration during sleep.

DISCUSSION

The dental and medical findings of both rickets and osteopetrosis have been well-documented.[1,3,4,6,7,12,15] When Rickets disease is associated with osteopetrosis, it is called osteopetrorickets, and the findings of osteopetrorickets have been defined as the mixture of findings of both rickets and osteopetrosis.[2,5,16,17] Previous publications have also reported that patients with osteopetrorickets often receive a primary diagnosis of rickets, based on clinical and radiographic findings. Only advanced examinations can reveal the symptoms of osteopetrosis, so that a final diagnosis of the actual disorder, osteopetrorickets can be made.[2,5] Our patient was also first diagnosed as rickets. Only radiographic investigation of the pathognomonic findings of osteopetrosis led to the accurate diagnosis of osteopetrorickets.

Bone marrow transplantation is a common medical intervention in the treatment of osteopetrorickets.[2,5,16,17] To achieve better results, however, the existing rickets needs to be treated completely before bone morrow transplantation. Likewise, our patient had undergone medical treatment for rickets before bone morrow transplantation. The destructive effects of osteopetrosis on teeth can be prevented if bone marrow transplantation is commenced as early as possible.18 In the present case, an appropriate (HLA-matched) donor was found when the patient was 2-years-old and thus the transplantation is expected to halt or minimize the effects of osteopetrosis on developing teeth thereafter.

The lack of osteoclast function in osteopetrosis leads to both severe sclerosis of the skeleton and disturbance of tooth eruption and periodontal tissues.[15,19] Delayed eruption, malformed teeth,

and enamel-dentin hypoplasia can be observed during the course of osteopetrosis. Osteopetrosis causes to thickening in lamina dura and defects in the periodontal membrane.[20] Foremost oral and dental findings of rickets are tooth malformation, multiple dental abscess without decay or trauma, and marked enamel-dentin hypoplasia.[4,6-8,14] It has been suggested that microorganisms are trapped within hypomineralised and malstructured dentin, causing pulpal infection and dental abscess.[21]

A search of the literature shows that dental and oral findings of osteopetrorickets have not been described in the dental literature previously. In the case presented herein, delayed eruption, enamel defects and hypoplasia, yellowish enamel and unerupted malformed teeth were observed. The erupted teeth were extremely mobile. These findings are, indeed, almost a mixture of those observed in osteopetrosis and in rickets. Delayed eruption, hypoplastic and hypocalcified dental hard tissue formation and malformed teeth are some of the oral and dental findings of either osteopetrosis or rickets. However, tooth mobility was not previously reported in rickets or in osteopetrosis. Because osteopetrorickets is a very rare disease and that we have been able to examine only one patient so far, we cannot claim tooth mobility to be a genuine dental finding of the disease.

Although osteomyelitis is the most important complication of tooth extraction in osteopetrosis patients, the mobile teeth of the present case had to be extracted, not only due to disturbances during the feeding but also for elimination of any potential means of focal infection before bone marrow transplantation. Subsequent osteomyelitis was not observed in periodical controls.

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