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CASE REPORT

Osteopetrosis: Oral and Maxillofacial Manifestations

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ABSTRACT

Osteopetrosis is a rare hereditary bone disease characterized by osteoclasts malfunction and impaired bone resorption. Decreased vascularity of bone as well as compromised immune system may result in oral and maxillofacial complications. Osteomyelitis is life-threatening problem in these patients usually associated with dental caries or abscess. Dental practitioners could play a crucial role in early diagnosis of osteomyelitis and avoid inappropriate treatments and further complications.

Keywords: Osteopetrosis, Dental complications, Dental managements.

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INTRODUCTION

Osteopetrosis (OP; Albers Schonberg disease, osteopetrosis generalizata, marble bone disease) is a rare hereditary bone disease characterized by osteoclasts malfunction and impaired bone resorption and consequently, a decrease in bone turnover and formation of immature bone.[1-4] The estimated incidence is 1 in 100,000 to 500,000.[5] This disease has been reported in three clinical forms: (1) Malignant infantile form with poor prognosis and autosomal recessive inheritance, (2) benign adult form with autosomal dominant inheritance and associated with few symptoms, (3) autosomal recessive intermediate form with clinical manifestations similar to malignant form and lowest incidence rate.[5-8] OP may associate with various symptoms.[3] Osteomyelitis is the common complication observed in osteopetrotic jaws following trauma.[2-4] Treatment of osteomyelitis in these patients is a big challenge for dental practitioners. We present a case of OP with osteomyelitis of the mandible with comprehensive review of disease.

CASE REPORT

A 23-year-old white male with known OP was referred by his family physician to the Pediatric Dental Clinic for management of left facial swelling. According to his medical records, mother's delivery was natural and his parents were far relatives and healthy with eight children without any history of OP. At the age of 8, he was hospitalized due to severe cardiac failure, hepatomegaly and severe splenomegaly. At 11 years of age, he suffered from severe visual disturbance and noticeable decrease in hearing.

Oral examination revealed reverse overjet at the anterior area and severe bilateral crossbite at the posterior region. Unerupted mandibular canines resulted in spacing between the anterior permanent teeth (Fig. 2). Periapical and cephalometric radiographs showed malformed teeth with no evidence of apical or periapical abscesses (Figs 1 and 3). The molar teeth had a sloped occlusal surface with malformed crown and short roots (Fig. 3).

At the age of 23, the patient was referred to us for management of left mandibular swelling and redness. The swelling had been developed over 2 weeks prior to admission, with left gingival pain. Mouth opening was restricted. Intraoral examination revealed recently extracted left mandibular first molar and purulent discharge in the gingivolabial sulcus at the left mandibular area. Facial examination showed erythematic swelling with draining

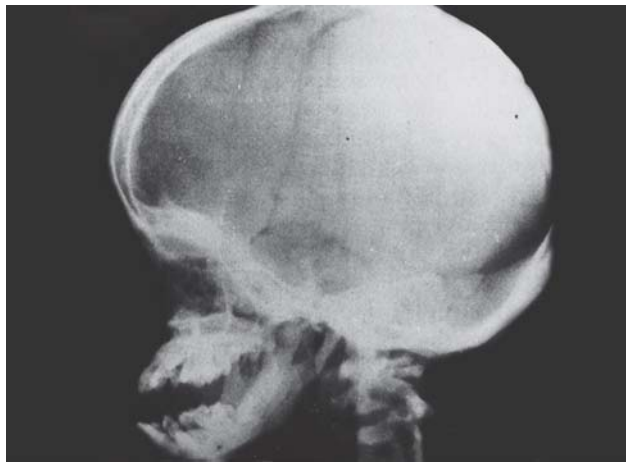

Fig. 1: Lateral cephalometric radiography of the patient

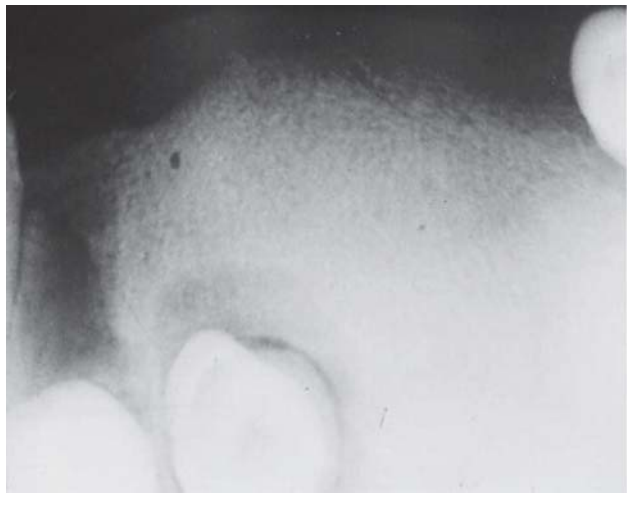

Fig. 2: Radiographic appearance of unerupted permanent teeth

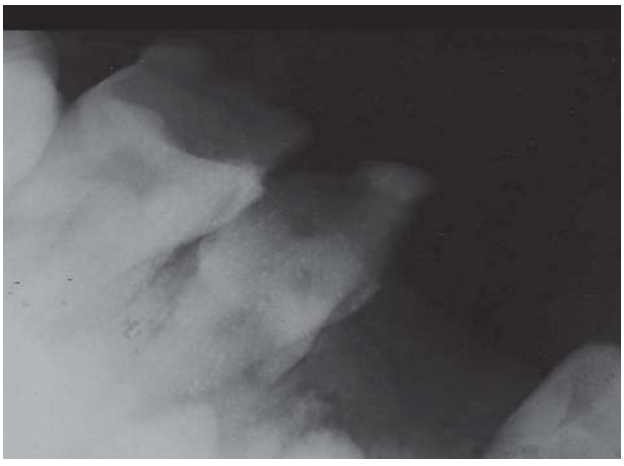

Fig. 3: Periapical radiograph shows teeth with malformed crown and short roots

fistula that progressed over left mid third of face. The head and neck examination revealed flattened malar eminence and dolicocephalic skull. A biopsy was performed and pathological report was consistent with osteomyelitis. After bacterial culture, therapeutic approach included broad spectrum antibiotic therapy, drainage of abscess, debridement of necrotic tissues and sequestrum, and daily irrigation of affected area with iodine. The chronic osteomyelitis was not treated completely and remained for many years. Acute phases of osteomyelitis were managed with intravenous antibiotic therapy. Although he suffered from severe visual disturbance and deafness, he received his bachelor degree in historical science successfully.

DISCUSSION

OP was first reported in 1904 by the German radiologist Albers-Schönberg.[1] The term osteopetrosis was described by Karshner in 1926.[9]

Depending on the form and stage of the disease, OP presents with variable clinical manifestations. Autosomal recessive OP or malignant infantile form of disease is a life-threatening condition usually initiated with anemia and hepatosplenomegaly in the first few months of life.[12] These patients are more susceptible to fractures and infections. Craniofacial skeletal changes can result in frontal bossing, hydrocephalus, macrocephaly, facial deformity, and choanal stenosis.[5,6] Delay in treatment will result in sepsis, pneumonia, hemorrhage and death.[10,11] Cranial nerve foramina may be narrowed by excessive development of cranial bones, resulting in deafness, visual disturbance and facial palsy.[10,13] Malformation of paranasal sinuses occurs due to impaired resorption and remodeling process, resulting in stuffy nose. The increased bone density affects the tooth morphology and inhibits tooth eruption.[5,12]

Autosomal dominant OP is the adult form of disease leading to osteomyelitis, fractures and sclerosis of the axial skeleton by affecting skeletal structure.[5,10] This form of disease is benign and asymptomatic in about 40% of patients.[14] Pressure on cranial nerves and bone marrow failure are rare.[5,10,14] Patients with autosomal dominant OP are susceptible to dental caries due to compromised hydroxyapatite crystal formation. Osteomyelitis is life-threatening problem in these patients usually associated with dental caries or abscess.[5,14] Two major radiologic signs of this disorder are diffuse increased radiopacity of medullary bone and sandwich vertebrae or rugger jersey appearance due to sclerosis of the axial skeleton.[15]

Autosomal recessive intermediate form of OP is milder than malignant condition with significantly lower rate of occurrence. Patients are usually asymptomatic with only radiographic signs.[16] This form of OP mimics orthopedic and orofacial symptoms of malignant disease such as nerve compression, macrocephaly, osteomyelitis, reduced arm span and several bone fractures. Ankylosed teeth might be seen in these patients making them susceptible to osteomyelitis of the jaw.[5,17] Hepatosplenomegaly and bone marrow failure are not frequently reported.[5,6]

Bone fractures are commonly reported in femoral shaft followed by the inferior neck of femor and posterior tibia.[15,18] Increased density of skull bones affects anterior cranial fossa more than other parts. Upon radiographic analysis, patients have poorly pneumatized sphenoid, mastoid and frontal bones.[19] Endobone or bone within bone may be seen in radiographs of hand, pelvis and scapula. Diffuse increased radiopacity of medullary bone is most routinely seen in dental radiographs.[5,15,19] Decreased vascularity of bone as well as compromised immune system make these patients susceptible to osteomyelitis of the jaw. Osteomyelitis of the mandible is more frequently reported in the literature and usually associated with tooth extraction in dental practice.[1,20]

Cummings et al[21] reported severe visual disturbance in a 6-month-old infant due to optic nerve compression. Our patient began to gradually suffer from blindness and deafness when he was 11 years old. In the current case, dental radiographs showed unerupted mandibular canines and malformation of other teeth.

Dental complications of OP are enamel hypoplasia, teeth with malformed crown and short roots, embedded teeth, poor oral hygiene and dental caries.[20] Constriction of inferior alveolar canal and dental pulp canal as well as thickening of the lamina dura are routinely seen in dental radiographs. In dental radiographs, bone marrow cavities and dental pulp chambers are not clearly recognized due to increased bone density.[1,22,23]

Bjorvatn et al[24] demonstrated severe malocclusion of primary and permanent dentition with significant decrease

in vertical growth of alveolar ridge. Our patient showed similar occlusion pattern with severe overjet at the anterior region and severe bilateral crossbite at the posterior region. The main odontogenic features in their patients were malformation of enamel, embedded teeth and dentinal abnormalities, all of which were observed in our patient.[24]

Management of osteomyelitis in patients with OP is controversial. Main therapeutic approach is antibiotic therapy combined with complete debridement of nectrotic tissue and suturing of soft tissue.[2-4] Hyperbaric oxygen therapy has been recommended for management of chronic osteomyelitis.[3,4] Higher oxygen content induces angiogenesis, promotes neutrophil-mediated defense and stimulates collagen formation.[3,4,22]

Preventive management with improved dental hygiene is the best therapeutic approach for patients with OP. Noninvasive root canal therapy is preferred to surgical procedures. As a general role, treatments should be performed as conservative as possible.[23, 25]

In the present case, osteomyelitis was initiated after extraction of mandibular molar. Debridement of necrotic tissue in combination with antibiotic therapy was performed for management of chronic osteomyelitis of the mandible. As the facility of hyperbaric oxygen therapy was not available in our setup, this therapeutic approach was not applied for our patient. Treatment of OP is multidisciplinary due to wide range of clinical symptoms. Dental practitioners could play a crucial role in early diagnosis of osteomyelitis and avoid inappropriate treatments and further complications.

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