Infantile osteopetrosis: a case report with osteomyolitis of the maxilla

Jose Martin Toranzo Fernandez* / Miguel Angel Noyola Frias** / Sandra Patricia Duarte Hernandez***

A case of a pediatric patient diagnosed with osteopetrosis complicated by osteomyelitis in the maxilla is presented. The combined medical-surgical treatment for this type of patient is discussed.
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INTRODUCTION

Osteopetrosis is a rare hereditary bone condition known as Albers-Schonberg disease or marble bone disease. Infantile malignant osteopetrosis is autosomal recessive and is characterized by skeletal sclerosis. It is associated with multiple fractures, anemia, extramedulary hematopoiesis, hepatoesplenomegaly and neurological abnormalites; like facial paralysis, blindness, deafness and pain from cranial nerve compression.[1-3]

The first case was reported in 1904 by Alberts-Schonberg in which he described a generalized skeletal sclerosis. In 1921 the German literature described six cases in which a peculiar radiographic appearance was seen resembling marble thus it was referred to as 'marble bones'. Later, the term osteopetrosis was used to described the principle pathologic alteration of the disease "petrifaction".[2,5]

This disorder is characterized by a defect in the osteoctastic function, resulting in the accumulation of sclerotic bone. The osteoclastic equilibrium is altered and bone apposition continues without normal physiological resorption leading to accumulation of bone in foramina and cranial nerve compression. This deficit results in abnormal bone remodeling, leukoerythroblastic anemia, immune dysfunction, alteration in vision and auditory changes.[1] Many patients exhibit stunted growth and development.

Described dental alterations in osteopetrosis include: late tooth eruption, congenitally missing teeth, malformed teeth, enamel hypoplasia, abnormal pulp chambers, early exfoliation of primary teeth, periodontal ligament defects and caries.

Prognosis is poor with death usually occurring prior to the age of 20. The most common cause of death is related to severe chronic anemia, bleeding or infection.[8] Osteomyelitis of the jaws may occur after odontogenic infections or surgical dental procedures. Osteomyelitis is a serious life-threatening sequella that is seen in the most frequently in the mandible followed by the maxilla, scapula and extremities.[4,5]

A case of a pediatric patient diagnosed with osteopetrosis complicated by osteomyelitis in the maxilla is presented. The combined medical-surgical treatment for this type of patient is discussed.

CASE REPORT

A 9 year old female was referred to the Oral and Maxillofacial Surgery Service for evaluation and treatment of a swelling in the left maxilla, which had been present and increasing in size over the past 10 months.

Examination revealed facial swelling over the left zygomatic-maxillary complex approximately 2.5 x 3cm in diameter. The area was erythmatous and painful to palpation. A sinus tract was present on the overlying skin that was draining purulent material (Figure 1). Additional examination revealed generalized pallor, parietal cranial prominence and exopthalmos. Intraoral exophitic lesions were noted in the left buccal region (Figure 2). There was a left palatal expansion with erythmatous overlying mucosa. There

* Dr. Jose Martin Toranzo Fernandez, Chief Oral and Maxillofacial Service, Hospital Central ' Dr. Ignacio Morones Prieto', Universidad Autonoma de San Luis Potosi.
** Dr. Miguel Angel Noyola Frias, Assistant Professor, Department of Oral and Maxillofacial Surgery, Hospital Central Dr. lgnacio Morones Prieto'.
*** Dra. Sandra Patricia Duarte Hernandez, Resident Oral and maxillofacial Surgery, Hospital Central 'Dr. lgnacio Morones Prieto

Send all correspondence to Dr. Jose Martin Toranzo Fernandez, Chief Oral and Maxillofacial Service, Hospital Central ' Dr. Ignacio Morones Prieto', Universidad Autonoma de San Luis Potosi, Av. Venustiano Carranza #2395, Colonia Universitaria C.P. 78240, San Luis Potosi, S.L.P., Mexico.

Voice: 01(8) 13-03-43 01(8) 14-64-74
E-mail: sdhcmf@hotmail.com.mx

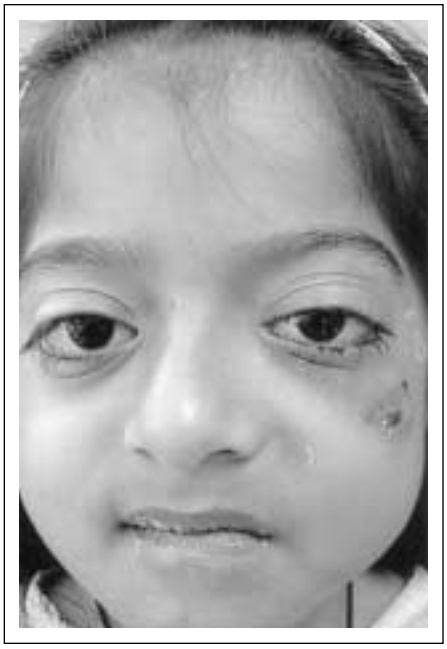


Figure 1. Front view showing swelling in the left zygomaticomaxillary complex.

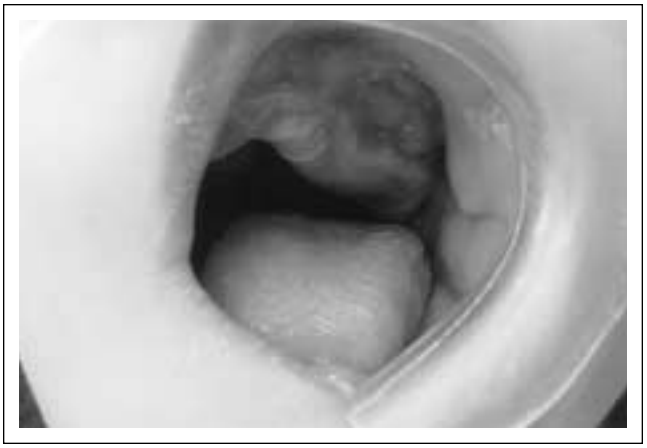
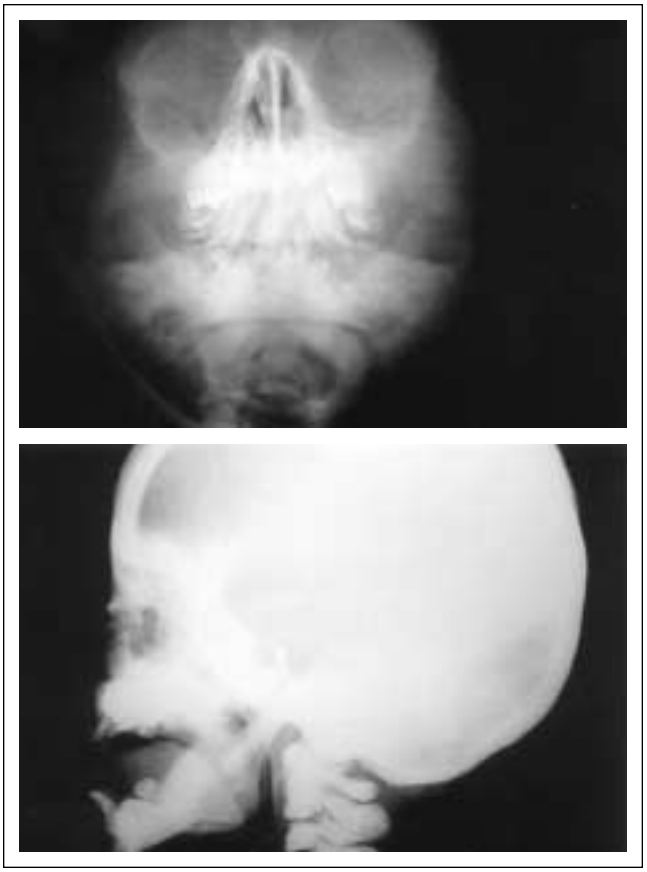


Figure 2. Intraoral photograph showing exophitic lesions in the left buccal region.



Figures 3, 4. Lateral and front view of cranial bone of jaws is extremely dense.

were no erupted teeth in the maxilla and the mandible exhibited primary anterior teeth.

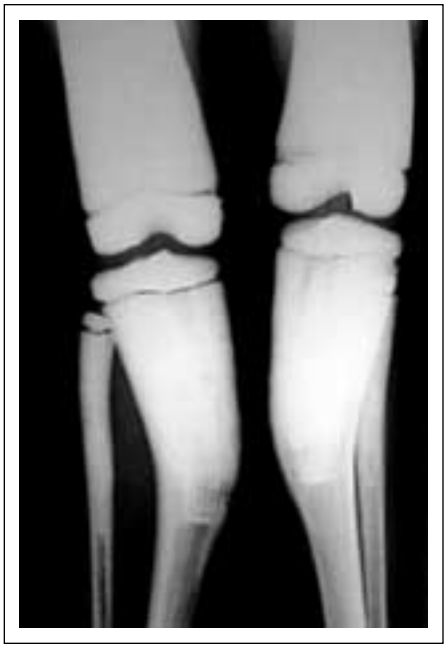
Review of the past medical history revealed the diagnosis of osteopetrosis. The diagnosis was made at the age of seven when the patient presented with scoliosis that limited movement, and was thought to be delaying normal growth and development. A radiographic and metabolic work-up lead to the diagnosis of osteopetrosis. There was no history of osteopetrosis or related neurological and hematological alterations in the family.

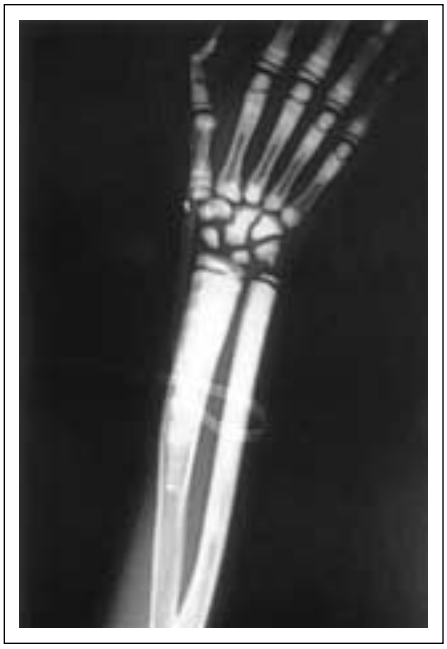
The symptomatology started nine months prior to presentation with a small swelling over the left malar area accompanied by acute dental pain. The patient initially sought care from her general dentist, who rendered treatment, which resulted in some resolution of symptom (specific treatment is unknown). Over the next 8 months the patient and her parents report increased pain, and swelling that was treated with over-the-counter medications without seeking medical attention.

At the time of presentation the patient was noted to be short in stature and weighed only 39 lb. Skeletal deformities included: genu valgum, parietal-occipital bossing, and faces resembling Crouzan's syndrome (Figure 1). Her vision was found to be slightly diminished with decreased peripheral visual fields and her hearing acuity tested normal. Radiographs of the chest, cranium and long bones revealed markedly increased radio-opacity (Figures 3 to 6).

The panoramic radiograph revealed sclerosis and increased density of the maxilla and mandible. There was multiple unerupted permanent teeth, malformed teeth, hypoplasia of the enamel and abnormal pulpal chambers (Figure 7). The left maxillary first and second molars had a radiolucency associated with them.

The laboratory studies showed anemia (hemoglobin of 6.12 g/dl, hct 19.8%), leukopenia 4900, alkaline phosphate 221 u/L, and remaining parameters normal.





Figures 5, 6. Radiographs of the long bones with markedly increased radiopacity

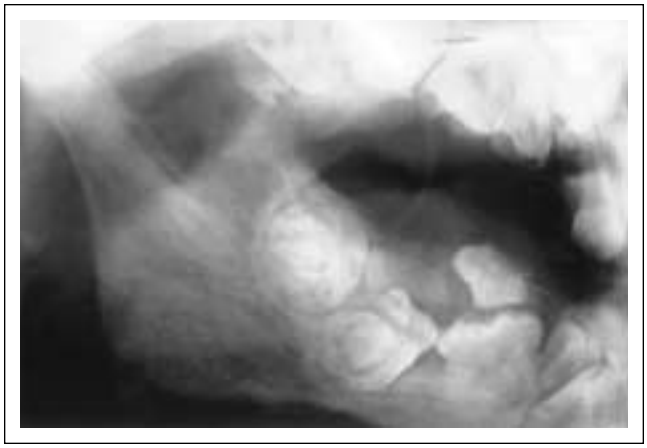


Figure 7. View of mandible showing extreme density of the bone and unerupted permanent teeth, malformed teeth, hypoplasia of the enamel and abnormal pulp chambers

Figure 8. During the surgical procedure the maxillary bone appeared petrified and avascular with without formation of sequestra

The initial therapeutic management consisted of: 1) empirical antibiotic therapy (Clindamycin 180 mg IV every six hours) accompanied with culture and sensitivities of the purulent debris, 2) correction of anemia (two units PRBC's) and 3) conservative surgical treatment, which consisted of curettage and debridement. During the surgical procedure the maxillary bone appeared petrified and avascular with/without formulation of sequestra (Figure 8). The bone and overlying soft tissue were biopsied and reported with osteomyelitis. Over the next 48 hours the patient showed marked improvement with decreased pain, swelling and correction of her anemia. Long term follow-up revealed the patient to be free of presenting symptoms and normal laboratory values.

DISCUSSION

Osteomyelitis is an inflammatory condition of the bone that starts in the bone marrow haversian systems and can extend to involve the cortex and periosteum. In general it is divided in two large groups: suppurative and nonsuppurative.[7] It is well recognized that conditions that alter bone vascularity and compromise circulation may increase the risk of developing osteomyelitis. Described treatment modalities of chronic osteomyelitis include: antibiotic therapy, surgical management, hyperbaric oxygen and immunotherapy. The conservative surgical treatment goes from incision and drainage, sequestrectomy, decorticating, resection and reconstruction when necessary. Treatment of osteopetrosis is aimed at decreasing or arresting progressive hyperostosis, cor-

recting anemia and thrombocytopenia, and treating infections. The oral cellulose phosphate, prednisone, low calcium diet, high dose of Calcitrol and recombinant human interferon gamma has been reported to be effective in some, but not all patients.[1,10] Bone marrow transplant has been reported to be curative in several osteopetrosis cases.[9]

This case of treatment of osteopetrosis complicated by osteomyolitis is reported as it responded very well to surgical debridement and medical management. Variable and mostly transient responses have been observed after treatment with intravenous parathyroid hormone, high dose calcitrol or prednisone. Recently, improvement of osteoclast function and abnormal clinical finding has been reported after recombinant human interferon gamma administration.[10]

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