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OSTEOPETROSIS

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(Report of a case and review of the literature)

Abstract

Osteopetrosis (Albers-Schonberg disease) is a rare disease characterized by generaized sclerosis of bones, hepatoslenomegaly, pancytopenia, multiple fracture, blindness.

The disease shows characteristic radiographic feature and two rather well-defined patterns have been recognized.

We experienced a case of osteopetrosis in a 31 year old man, who has had pus discharge and fetid odor after extraction of upper maxillary molar.

We made a brief review of literature.

INTRODUCTION

Osteopetrosis (Albers-Schönberg disease) is a rare generalized disorder of bone, characterized by a marked increase in density of the skeletal tissue[10].

In 1904 Albers-Schönberg first described the radiological appearance of marble bones. His first patient was a 26-year-old man, leading an apparently normal life, who displayed the symptom complex of generalized sclerosis of bone, anemia, hepatosplenomegaly, and multiple fractures. More than 200 cases have been reported since 1904 when Albers-Schönberg recognized osteopetrosis as a separate clinical entity[11].

It has become customary to place these cases into one of the two groups;

(1) a malignant form, which seen either prepatum or at very early age and progresses to a fatal termination rather early, or (2) a benign form, which often unrecognized. In many cases the latter type is compatible with a normal life expectancy.

The authors observed a 31-year-old male patient with osteopetrosis who had come to our Department of Dentistry, due to the complaints of pus discharge and halitosis on the upper jaw.

CASE REPORT

A 31-year-old male patient was admitted to our Department of Dentistry, Korea University on Oct. 27. 1988, for pus discharge and foul odor after extraction of left upper maxillary molar. On physical examination, heigt, physical development and proportions were normal. Mental retardation was not observed. He had a pale and anemic appearance with widened nasal bridge (Fig. 1-1).

He had also progressive blindness which had been worse and gait disturbance due to left femur fracture. The spleen was 17cm. Slight facial asymmetry was obvious as a result of swelling of the tissue overlying the left maxilla area. Mouth opening was restricted to approximately 3.5cm, measured between the incisal edge of the teeth.

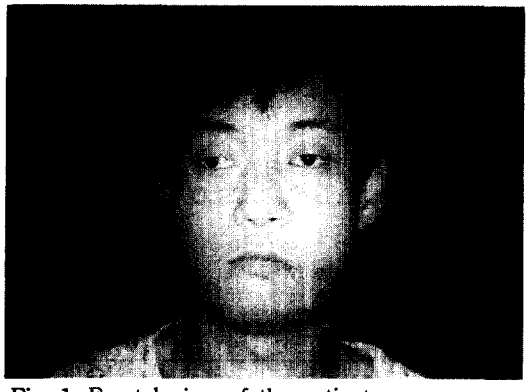

Fig. 1. Frontal view of the patient

Intraoral examination showed the general condition of mouth to be one of gross neglect, with many enamel dysplasia and broken-down teeth. In the left 1st molar region, the gingival tissue were severely recessed. A mass of necrotic bone occupied the left maxillary alveolar ridge in 2nd molar region. The necrotic mass was bathed in a purulent exudate.

Laboratory studies were the following:

Red blood count - 1330000/c. mm of blood

Hemoglobin - 3.8Gm/100ml of blood

White blood count - 3100/c. mm of blood

Platelet count - 4000/c. mm of blood

Acid phosphatase - 5.8 units

Urinalysis - normal

A full skeletal roentgenographic survey revealed sclerosis of all the bones of the body, including those of the skull and face (Fig. 1-2). Cortical bone could not be recognized since the marrow cavity appeared equally dense. Marked deformity of femur neck with multiple old fracture in left femur shaft (Fig. 1-3).

Dense sclerotic changes without normal trabecular in both femur, pelvic bone, proximal tibiofibular were observed, and the pulp chambers were not obliterated. The roots of teeth were generally small and dwarfed, and the alveolar bone appeared very dense (Fig. 1-4).

The hospital admissions for oral infection were due to complication after extration of upper left 2nd molar resulting in chronic suppurative lesions of the alveolar bone and periodontal tissue.

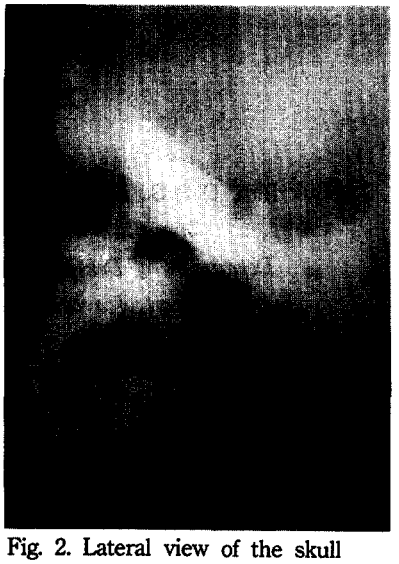

Fig. 2. Lateral view of the skull

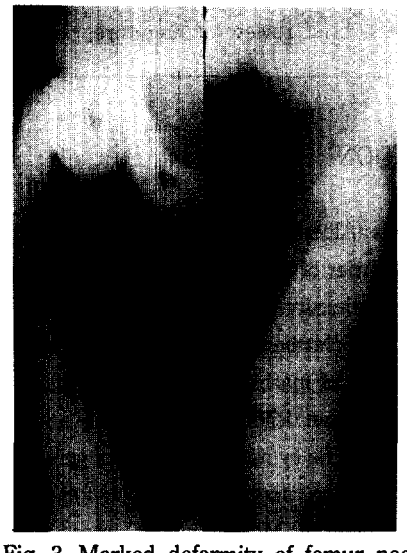

Fig. 3. Marked deformity of femur neek

Treatment consisted of antibiotics and anti-inflammatory durg coverage in addition to administration of platelet-rich plasma and packed RBC.

Sequestrectomy was done in upper left 1st. 2nd. molar region under local anesthesia (Fig. 5), and antibiotic therapy had to be continued for two weeks before normal healing commenced.

Stitch out was done at seventh day after operation.

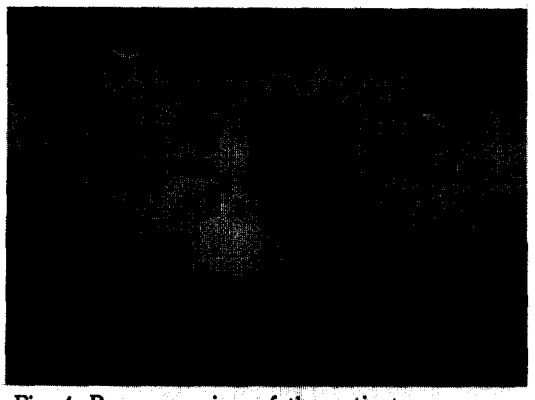

Fig. 4. Panorama view of the patient

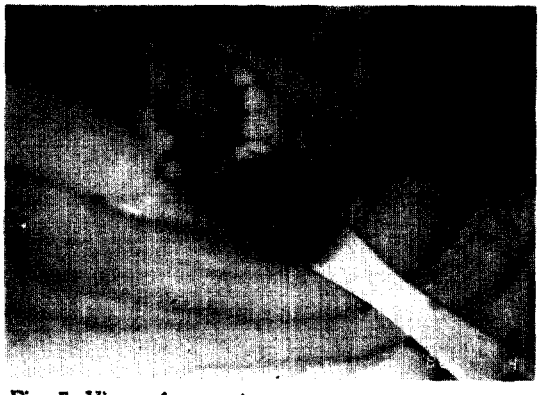

Fig. 5. View of operation

He discharged himself on Nov. 8. 1988. On March, 1988, the patient returned complaining of bony exposure on operation site. The same procedures were carried out, but the prognosis was not good.

REVIEW OF THE LITERATURE

Osteopetrosis, a rare disorder characterized by hardness and brittleness of the bones[10], has a variety of interesting medical and dental manifestation[11].

With progression of the disease, the bone become increasingly sclerotic and opaque to roentgenogram, the latter feature giving rise to the term marble bone. The change in bone structure is accompanied by a marked tendency toward fragility, and fractures may be sustained in even quite trivial accidents[10].

1) Clinical Features

The disease varies remarkably in its severity and speed of onset. Two rather well-defined patterns of the disorder have been recognized. The malignant form of the disease is characterized by serious sequelae, such as optic atropy, deafness, hydrocephaly, multiple fractures, facial nerve palsies, disturbances in growth of skeleton, and disturbances in dental development.

Hepatosplenomegaly with an accompanying anemia, leukopenia, and thrombocytopenia are often part of the fulminating form of the disease[9]. Therefore, these patients are often afflicted with osteomyelitis and chronic infections, some of which may ultimately prove fatal.

The benign form of disease may also be characterized by visual disturbances, progressive deafness, facial paralysis, abnormal dental development, and multiple fractures. The anemia and hepatosplenomegaly are seen often in this group. Involvement of hematopoietic organs is one of the primary clinical prognostications of the severity of the disease[2]. In general, though, the earlier in life symptoms appears, the graver is the prognosis.

A characteristic facial apperance has been described. The face is broad, with eyes far apart, the nose is flattened, and there is bossing of the frontal prominences[10, 13, 14].

They tend to fracture varies quite widely[10]. Incomplete fractures may occur and their existence may be unknown until accidently discovered during roentgenographic examination. They tend to be appear wherever the bone is stressed: common sites are the underside of the femoral heads and the upper ends of the tibial condyles where there is a concentration of weight-bearing stress. A number of reason for the fragility have been proposed, but it is generally agreed that the fault lies in the microstructure of the bone rather than its chemical composition[14].

Lack of mature lamella bone, a deficiency of collagenous fibrills, the presence of tracts, the occurrence

of outbursts of localized resorption, and the absence of a functional arrangement of the trabecular have been implicated.

This disease has been seen in many races, with no predilection for either sexes.

The possibility that a hereditory factor is a cause of this disease is strengthened by the fact that cases have been reported in successive generation and also by the fact that consanguity is reported in approximately 20 per cent of the cases[16].

There is a familial history of the disease in 40 per cent of diagnosed cases. It is thought to be a reccessive genetic trait in some families, with intermarriage increasing the likelihood of its occurrence.

2) Roentgenographic Finding

Roentgenography provides the principal means of diagnosis[16]. Roentgenographic changes consist of bilateral, symmetrical sclerosis of all bones, with gradual obliteration of the marrow space[9].

In the long bones the first change is seen as an increase in density of diaphysis, where there is progressive intrusion of cortex on the spongiosa without an overvall increase in the size of bone.

In many cases, vertical striation of increased density can be seen near the end of long bones, and some show transverse line of alternating radiolucency and radiopacity. The vertical striations are thought to represent calcification of columns of cartilage which are often found in these abnormal bones, and the transverse bands represent alternating periods of remission or exacerbation of the process.[16].

A sclerotic area may be seen in the bones of the hand, where it appeares as a nidus of bone with a less dense periphery. Hin fel and Beiler refer to this as a "bone within bone" appearance.

The clubbing deformity of the long bones which consistently occurs in the malignant osteopetrosis is not a feature, but there may be some increase in diameter of the shafts of the long bones, particulary in the distal half of the femur[5]. The bodies of the vertebrae show a translucent zone sandwiched horizontally between zones of sclerosis[10].

In the skull, the greatest degree of density is found in the base. The posterior clinoid processes are thickened and club-shaped when seen on lateral skull roentgenograms. Because of bony encroachment upon the pituitary gland and cranial nerves it may lead to blindness, deafness, and diabetes insipidus.

The facial bones become extremely radiopaque and the paranasal sinuses are obliterated.

The deciduous and permanent dentitions are frequently malformed, with oligodontia, hypoplasia, or delayed eruption of teeth as common findings.

3) Histologic Finding

At present, the most plausible is the suggestion that the disease is a developmental error of faulty differentiation of the primitive mesenchyme which is the precursor of the osseous blastoma.

It is postulated that osteogenesis is disturbed, with persistence and proliferation of poorly differentiated osteoid tissue which resist resorption.

The microscopic picture varies widely depending on the stage of the disease, the age of patient, and the region examined. In general, osteopetrosis is manifested by excessive deposition of highly mineralized tissue in both cortex and the spongiosa. There is persistence of calcified cartilagenous and osteoid elements, with a marked deficiency of collagenous fibrils and a conspicuous paucity of mature lamellar bone.

Haversian systems are irregular or missing, and osteocytes may be spindle-shaped, infrequent, or absent.

In area of severe sclerosis, only necrotic bone may be seen. Some have reported osteoblasts in normal numbers, but osteoclast may be deficient[6] or of unusual size with large numbers of nuclei[12].

CONCLUSIONS

Osteopetrosis display the generalized sclerosis of

bones, and the deciduous and permanent dentitions are frequently malformed. The cause of death is chiefly anemia by excessive hemorrhage and sepsis by infection.

Because the patients with osteopetrosis have high susceptibility to dental caries, and the resistance to infection is decreased with a tendency toward osteomyelitis after oral and maxillofacial surgery, it is essential that one should take a medical history for patient and make provision against complication of dental treatment.

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골석화증에 대한 문헌고찰

국문초록

Osteopetrosis는 드물게 볼 수 있는 질환으로 일명, Albers-Schönberg disease 혹은 Marble bone disease라고도 하며 그 주요한 특징은 전반적인 골경화증, 임파선의 증대, 간비 증대, 범혈구 감소증, 다발성 골절과 실명 등이다.

이 질환은 X-선상에서 독특한 소견을 보이며 임상적으로 악성형과 양성형으로 나눌 수 있다.

Dominant form은 양성형으로써 다양하게 표현되어 단독 X-선상에서 이상을 나타내는 것부터 골절과 뇌신경을 침범하기까지 다양하게 나타나며, Recessive form은 악성형으로 자궁내에서 진단되어지고 빈혈이 심하며 골수양화생이 있으며, 감염 특히 하악의 골수염으로 대개는 첫 1년 이내에 사망 한다고 보고되어 있다.

이에 저자들은 개인치과에서 상악 좌측 제2대구치 발거후 골수염이 발생한 31세 남자 골석화증 환자를 치험하였기에 문헌고찰과 함께 보고하는 바이다.