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Alberg's Schoenberg Disease – A Review and Case Report

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ABSTRACT

Osteopetrosis is a rare hereditary bone disease characterized by a abnormal overgrowth of dense bone because of osteoclasts malfunction, impaired bone resorption, decrease in the rate of bone turnover and formation of immature bone. There are a number of clinical variants with significantly different prognosis and clinical behaviours that have been described in the literature. The diagnosis of Osteopetrosis is largely based on the clinical and radiographic evaluation, and confirmed by gene testing. Treatment for osteopetrosis is largely symptomatic, however haematopoietic stem cell transplantation is currently the best chance for the long term survival of the patient. Osteomyelitis is a known complication of osteopetrosis. A prompt diagnosis, appropriate treatment and a continuous follow up decreases the long-term sequelae of the disease. This article emphasis on the signs and symptoms, clinical and radiographic features of Osteopetrosis and gives an insight on a case diagnosed with Osteopetrosis in an eight year old female patient and about the treatment rendered.

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I. INTRODUCTION

The term osteopetrosis is derived from the Greek word 'osteo,' meaning bone, and 'Petros,' meaning stone. It is also known as marble bone disease, osteosclerosis fragilis, osteitis condensans, Alberg's Schoenberg disease(1). The history of Osteopetrosis can be traced to 1904. The case Osteopetrosis is first documented by Heinrich Albers Schonberg, and Karshner coined the word "osteopetrosis" in 1926. (2)(3).

Osteopetrosis is a rare heritable genetic bone disorder is characterized by an increase in the cortical bone mass,as a result of defect in the osteoclastic function. The reported rate of incidence varies from 1:100 000 to 1:500 000 (4). 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)(6)

An interruption in the normal bone modeling and remodeling can give rise to skeletal deformity and dental abnormalities. Diagnosis is mainly based on clinical and radiographic evaluation, which is then confirmed by

genetic counseling. Osteopetrosis is characterised by a variety of systemic and oral complications. The treatment of osteopetrosis is based on the mutation and clinical comorbidities that requires a multidisciplinary team approach. The team includes specialists from endocrinology, ophthalmology, genetics, dentistry, orthopedic surgery, neurology, neurosurgery, otolaryngology, hematology, infectious disease, nephrology, pain management, and developmental paediatrics(7).
One of the major oral complication in osteopetrosis is osteomyelitis, particularly in the mandible.This paper illustrates about various forms of osteopetrosis and a case report on osteopetrosis of mandible.

II. CASE REPORT

An eight year old female patient a known case of osteopetrosis with a chief complaint of pain in left lower back tooth region for the past 1 week was referred to the Department of paediatric and Preventive Dentistry of Adhiparashakthi Dental College and Hospital by a neighbouring private hospital. On extra oral examination, the patient was found to be moderately pale with poor nutritional status, short stature, vision loss, frontal bossing and malformed craniofacial appearance. Intraorally there was no soft tissue abnormaility detected except for the color of the gingiva which was highly pigmented, there was a premature exfoliation of 54,55,65,73,83,84,85 and also was noted with U shaped alveolar arches.The patient complained of difficulty in mastication and speech. Patient was advised a digital orthopantamogram which revealed a generalized increase in bone density with absence of normal trabecular pattern involving both jaws, indistinct coronoid process with an increased gonial angle, Marrow spaces cannot be appreciated clearly, root resorption evident irt 74,75,ectopic eruption of teeth,Abnormal tooth structure and eruption sequence. Majority of permanent tooth follicles were absent. A generalised radiopacity of maxilla and mandible is evident with multiple unerupted tooth. She had been diagnosed with Autosomal recessive osteopetrosis with no relevant family history and Consanguinity. The patient was advised symptomatic and supportive treatment of Analgesic and Antibiotic therapy. Patient was also advised for extraction of 75 .An Antibiotic prophylaxis cover was advised to the patient to prevent the occurrence of osteomyelitis of mandible. Amoxycillin 250mg was given 1hr prior to the dental procedure. Informed Consent was obtained from the parents before performing the extraction.Topical anaesthesia was achieved with 2% lignocaine gel following which 2ml of 2% lignocaine with 1:2,00,000 adrenaline was administered using 2ml syringe. Subjective and objective symptoms were verified after which extraction was done irt 75. Post extraction socket was analysed for tooth remanants after which adequate haemostasis was achieved. Post extraction instructions and medications (Antibiotics and analgesic) was prescribed. Patient was under continuous follow up to prevent the occurrence of osteomyelitis and to ensure a proper oral health.

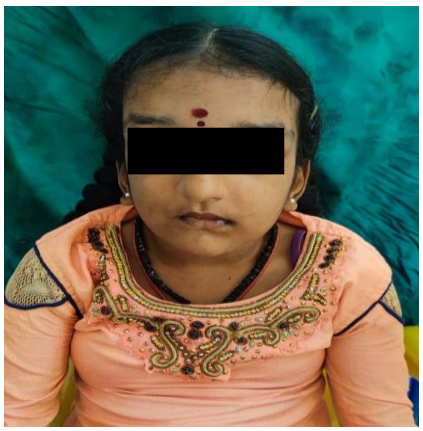

Figure 1
Figure 1:Extra oral profile showing malformed craniofacial appearance

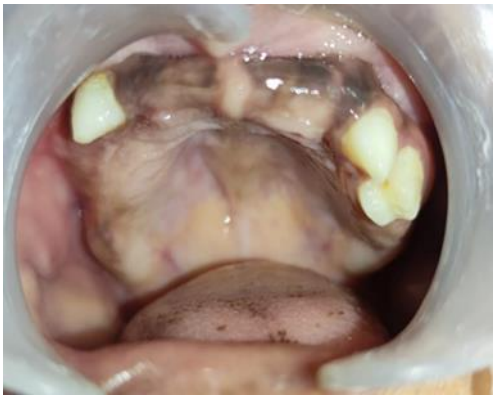

Figure 2

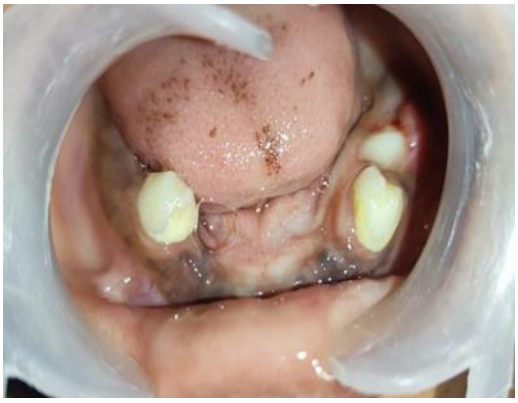

Figure 3

Figure 2 and Figure 3 illustates Intraoral maxillary and mandibular occulsal view which shows premature exfoliation of teeth in maxillary and mandibular arch.

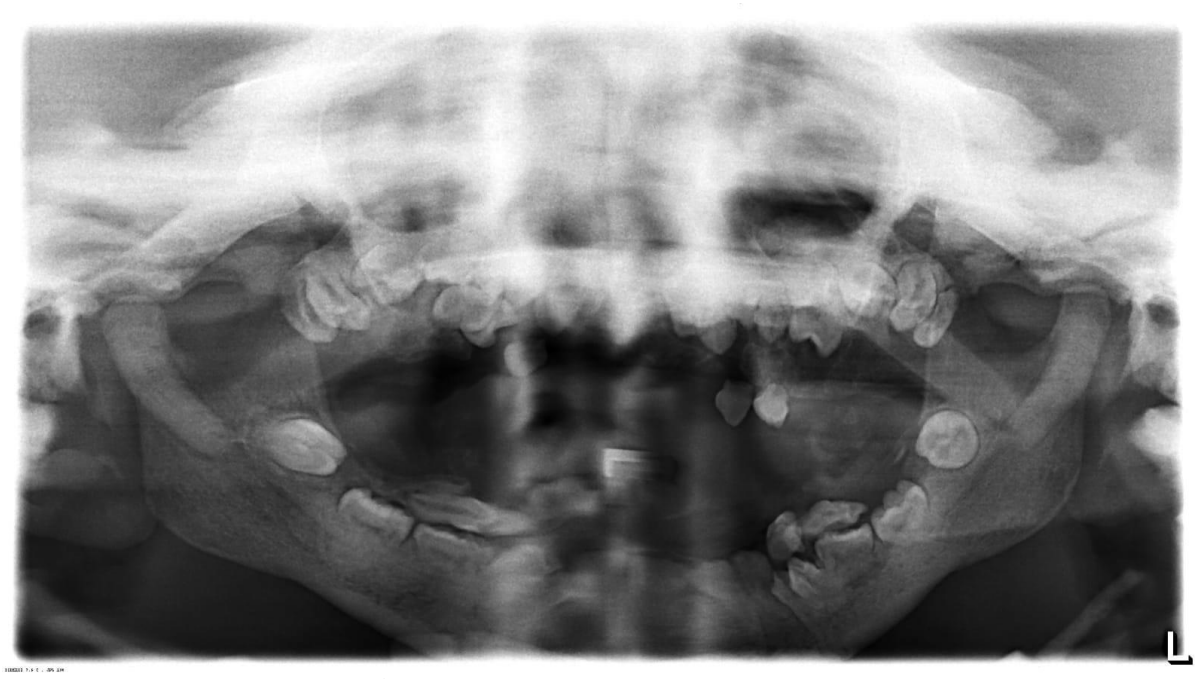

Figure 4

Figure 4: orthopantomogram reveals generalised increase in bone density with absence of normal trabecular pattern involving both jaws, root resorption evident irt 74,75, ectopic eruption of teeth, Abnormal tooth structure and eruption sequence.

Discussion:

Osteopetrosis is a genetic disorder characterized by mutations in CLCN7, TGIRG1 genes leads to a defective carbonic anhydrase II enzyme function in osteoclasts because of which there is a defective bone resorption leading to increase in bone density(2)(8).

There are 3 main types of osteopetrosis.

1. Autosomal recessive osteopetrosis (ARO)
2. Intermediate autosomal recessive osteopetrosis (IARO)
3. Autosomal dominant oseopetrosis (ADO).

Autosomal recessive Osteopetrosis which is also called as malignant or infantile osteopetrosis, is the most serious type. It is often diagnosed early after birth, and it is lethal without treatment. The estimated prevalence is 3 to 34 individuals per million(9,10)In ARO, bone remodeling is extremely inhibited, due to non-functional or scarce osteoclasts(2,8).Children suffer from anemia and immunodeficiency thus making them prone to high risk infectious diseases.They also suffer from visual impairment due to the growing bone pressing on the optic nerve; it mostly occurs in the child's first year after birth.In addition to visual impairment , approximately one third of children with ARO will have hearing loss. ossification of the cranium fontanels may begin early, making the brain not have sufficient room to grow, leading to occurrence of cognitive disabilities(10). Eye sockets are shallow which gives an appearance of protruded eyes.Flattened nasal bridge.Since the bone are more dense and fragile they are more prone to fractures.ARO leads to a variety of dental complications such as aplasia,delayed eruption in both primary and permanent dentition,little or no tooth enamel,misshaped dentin.They are also highly susceptible to the occurrence of osteomyelitis(11,12) Our case was diagnosed with the same where the child suffered from anemia,visual impairment,aplasia,delayed eruption in both primary and permanent dentition, and she also was more prone to repeated skeletal fractures she had a short stature, frontal bossing and malformed craniofacial appearance.
Intermediate autosomal recessive osteopetrosis (IARO):IARO is similar to ARO, IARO is seen during late infancy or early childhood, and the symptoms develop more slowly(2).The estimated prevalence of IARO is three individuals per million(2)Children genially have generally have enlarged spleen and liver.they also develop renal tubular acidosis and short bone lengths thus making them highly prone to bone fractures(3)Childern will have some degree of mental retardation. Hematopoietic Stem Cell Transplantation (HSCT) treatment is important for preventing disease progression and early death certain children survive to adulthood even without any treatment(9,10).

Autosomal dominant osteopetrosis (ADO):Children have mild symptoms, and some individuals do not experience any symptoms. It is seen in older children or young adults.ADO does not affect the lifespan of an individual.Patients have a classic sign of "bone in bone" appearance of the vertebrae giving rise to "sandwich" vertebrae and "rugger-jersey" spine(2). Children suffer from cranial nerve compression, anemia and immune insufficiency.oral complications are fractured tooth, multiple caries, mandibular osteomyelitis multiple dental abscesses, and mandibular fractures(8).
Diagnosis is mainly based on clinical,haematological and radiographic evaluation, which is then confirmed by genetic counseling. Radiograph aids to diagnose osteopetrosis are MRI, (HR-pQCT), DXA scan(9)(10)(13).The levels of serum calcium, phosphorous, alkaline phosphatase,acid phosphatase to be evaluated.Generally the levels of serum calcium, phosphorous, alkaline phosphatase are within normal limits with elevated acid phosphatase(11)
The systemic treatment for osteopetrosis include calcium, calcitriol, interferon gamma, corticosteroids, erythropoietin, and human stem cell transplant.
Generalised dental considerations in osteopetrosis patients are increase in bone density of the maxilla and mandible with abnormal tubercular pattern, delayed eruption in both primary and permanent dentition, aplasia, Loss of enamel, misshaped dentin, enlarged dental pulp, thickened lamina dura, malformed crowns and roots, poor calcification, more prone for caries, susceptible to fracture of tooth and jaws. They are also highly susceptible to the occurrence of caries, dental abscess formation and osteomyelitis of the jaws(15).
Dental extraction would be a very common therapeutic intervention in these patients because of the defective enamel formation, increase bone density, abnormal tooth morphology and poor oral hygiene. However, performing these extractions poses a great difficulty as there is a high chance of formation of necrotic bone that leads to a protracted osteomyelitis and also oro-cutaneous fistulae. The maxillary tooth erruption may lead to a severe infection like orbital cellulitis (14).
A routine dental checkup, preventive measures,continuous follow up is necessary to ensure a good oral hygiene inorder to prevent dental complications like osteomyelitis as Osteopetrosis patients are more prone to develop infections and jaw fractures, hence surgical procedures even tooth extraction should be considered as a last resort of treatment rendered(12).

III. CONCLUSION

Osteopetrosis is a heritable genetic disorder due to defect in osteoclasts formation characterised by dense formation of bone making them fragile,thus making them more prone to fracture of the jaw and long bones.To avoid any dental complications and to ensure good oral hygiene adequate preventive measures,regular follow ups have to be ensured in osteopetrosis patient.
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