Osteopetrosis and Osteomyelitis-Their Secret Bond

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Abstract

Background: Osteopetrosis, is characterized by a spectrum of conditions marked by increase in bone density due to defective osteoclast activity, leading to fragile bones, hematologic issues, nerve entrapment leading to growth challenges. On the other hand, osteomyelitis is an inflammatory process of bone and bone marrow caused by an infectious-organisms which results in local bone destruction, necrosis and apposition of new bone.

Case report: This article discusses a case of a 32-year-old male initially diagnosed with maxillary osteomyelitis who presented with a draining fistula in the posterior maxilla for seven months. After a thorough combination of history, clinical, radiographic, and laboratory findings, it was discovered that the patient also had osteopetrosis. The hidden connection between the two conditions suggests that osteopetrosis may often go undiagnosed in patients presenting with osteomyelitis.

Conclusion: This case underscores the diagnostic challenges posed by overlapping pathologies such as osteomyelitis and osteopetrosis, particularly within the craniofacial region. The co-occurrence revealed in this patient emphasizes the need for heightened clinical suspicion and a multidisciplinary approach when evaluating persistent maxillofacial infections. Early recognition of underlying osteopetrosis is essential for appropriate management and may prevent prolonged morbidity.

Keywords: Bone fractures, Maxillofacial disorders, Osteomyelitis, Osteopetrosis

Introduction

Osteopetrosis is a rare hereditary bone disorder that results in an increase in bone density due to gene mutations and osteoclastic dysfunction. The overall incidence of this condition is difficult to estimate but autosomal recessive osteopetrosis (ARO) has an incidence of 1 in 250,000 births, and autosomal dominant osteopetrosis (ADO) has an incidence of 1 in 20,000 births.[1] The disorder is classically divided into 3 types with variable clinical features ranging from neonatal onset with life-threatening complications such as bone marrow failure (*e.g.* classic or "malignant" ARO), to the incidental finding of Autosomal Dominant form osteopetrosis in adults on radiographs.

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Case Report

A 32-year-old male presented to the Department of Oral Medicine and Radiology with the chief complaint of recurrent swelling on the right side of his face accompanied by pus discharge in the maxillary posterior teeth region for the past six months. The patient also reported intermittent pus discharge with a foul taste in his mouth.

Past medical history revealed that the patient had sustained fractures of long bones twice following a fall from stairs eight years ago. Upon further inquiry on family history, the patient disclosed that his father had been diagnosed with a bone disorder several years ago although the specific nature of the disorder was unknown to him.

Upon intialexamination, the patient presented with notable dysmorphic features, including short stature, facial puffiness and frontal bossing. Additionally bilateral proptosis was observed.

Extra orally facialswelling on the right sidewas noted, which on palpation was tender, fluctuant and soft. [Figure 1].

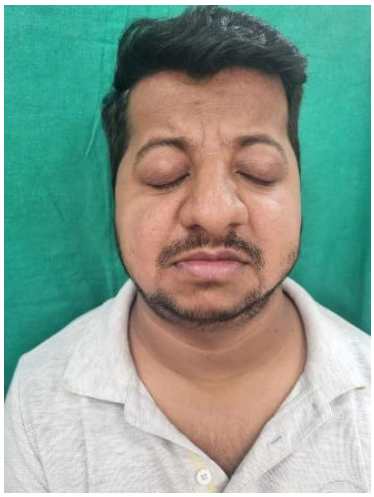

Figure 1: Showing diffuse extra oral swelling towards right side and puffy cheek appearance.

The first molar of the first quadrant was extracted about seven months back.

Intraorally, there was a break in the continuity of the oral epithelium, with partial edentulism, crowding and narrow constricted palatal arch.[Figure 2].

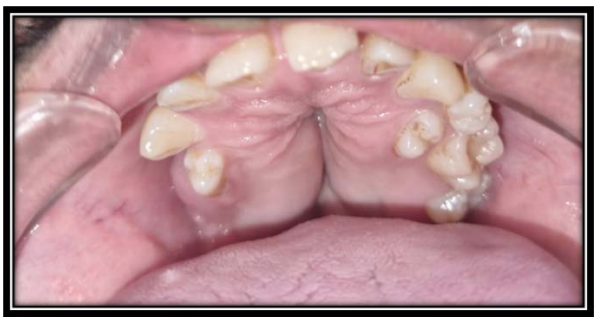

Figure 2: Missing 15, 16, 17, 18; palatally displaced 14, 24; bony exostoses B/L wrt mid palatine raphe, constricted arch and macroglossia.

Provisional diagnosis of oro-antral fistularelated to right maxillary first molar and differential diagnosis of osteomyelitis were made.

Radiographic Findings

- P.A. Water's View was done to view whether there was any oro-antral communication revealed haziness in frontal and maxillary sinus with dense sclerotic bone and thickened calvaria. [Figure 3].
- CBCT- Showed expansion and discontinuity of the buccal and palatal cortex, confirming the presence of osteomyelitis. [Figure 4].
- OPG and Lateral Cephalogram-Demonstrated a bone-within-bone appearance, generalized hypercementosis, hypoplastic mandible [Figure 5-A] open fontanelles, and hyperostotic vertebral end plates. [Figure 5- B].
- Blood investigations showed raised serum acid phosphatase.

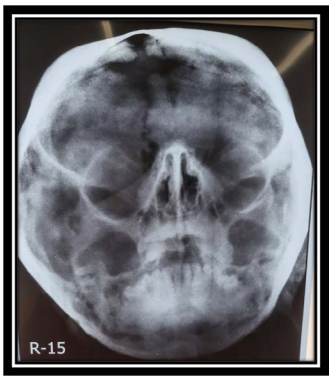

Figure 3: Waters view radiograph showing dense sclerotic marble bone appearance and thickened calvaria.

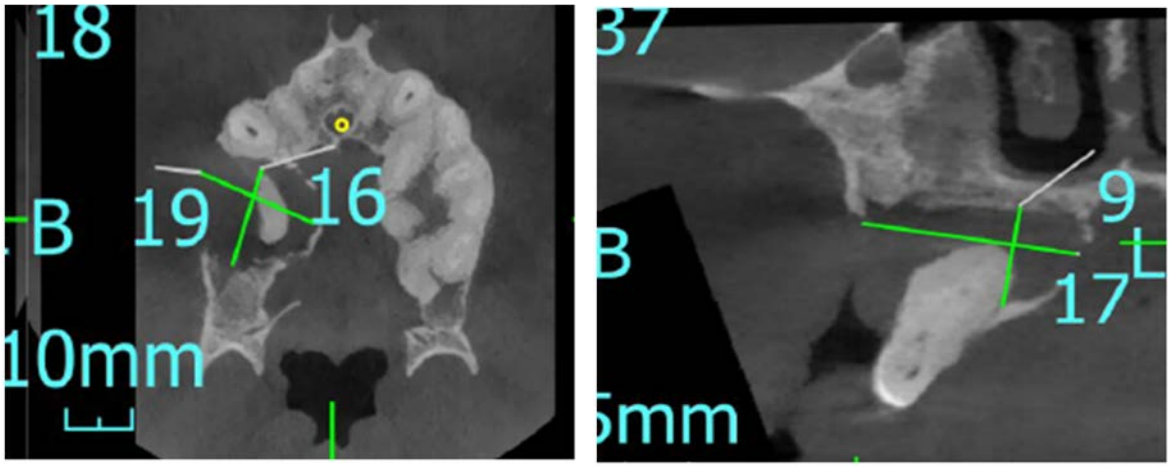

Figure 4: CBCT maxilla showing A) expansion of the palatal cortex B) break in the continuity in the buccal and palatal cortex.

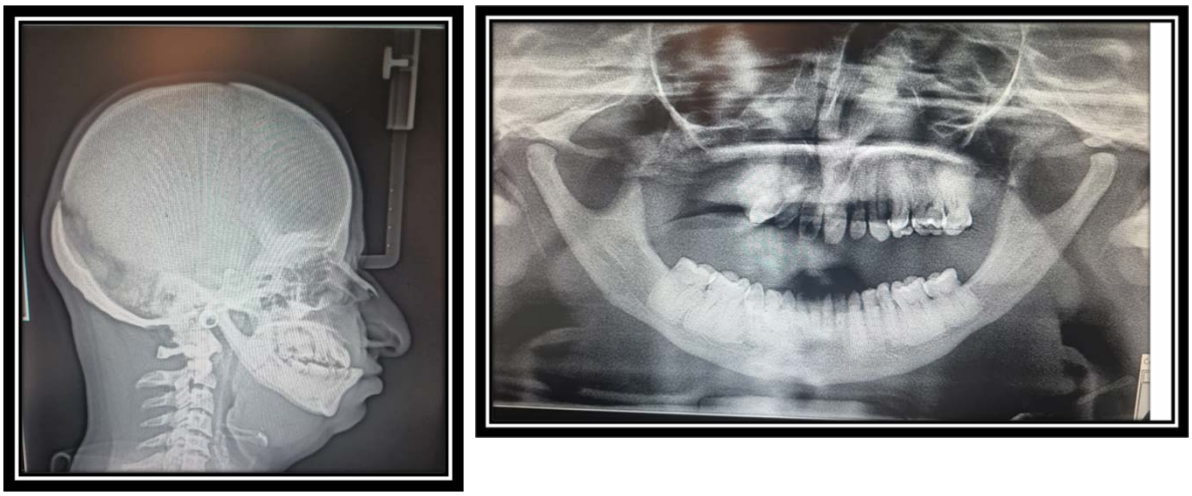

Figure 5: A) Lateral cephalogram showing open fontanelles, hypoplastic maxilla and hyperostotic vertebral end plates B) Orthopantomogram showing generalized hypercementosis, hypoplastic mandible and malformed TMJ complex

Based on history, clinical findings, and radiological examination, diagnosis of maxillary osteomyelitis with osteopetrosis was made.

Surgical closure of Oro antral fistula was done. The patient received a 7-day broad-spectrum antibiotic course (clindamycin 300 mg six hourly) and oral hygiene instructions. Orthopedic and oral surgeon consultations were sought for osteomyelitis management.

Discussion

Osteopetrosis is a rare hereditary disease caused by an imbalance in bone metabolism, resulting from decreased osteoclast activity. [2] Osteomyelitis is thus a well known complication associated with osteopetrosis due to its hypo vascular nature. [6]

Multiple genetic mutations are responsible for the presentation of osteopetrosis. The mutated genes of significance include CLCN7 and TGIRG1, which results in the functional defects of the enzyme carbonic anhydrase II. Osteoclasts ultimately cannot form ruffled borders, causing defective bone resorption with the subsequent accumulation of bone, thereby increasing bone density and the fracture risk.[3] This adversely affects the healing process and results in a marked

increase in infection susceptibility.As bone becomes denser, its marrow cavities and the pulpal chambers of the teeth become obliterated with the resultant constriction of the neurovascular bundles supplying the jaws and the teeth. [5]

The Autosomal Dominant Osteopetrosis Type II form, also known as Albers–Schonberg disease, is the most common and can have varied clinical presentation ranging from poor prognosis to being diagnosed unexpectedly from radiographs. [4]

A general rule suggests that a comprehensive history, combined with physical examinations, radiographic assessments, and supported by laboratory investigations, plays a crucial role in diagnosing underlying disorder.

Radiographic examination reveals bone within bone appearance in the cervical vertebrae, thickened calvarium, diffuse sclerosis, open fontanelles, sclerosis of skull, spine, pelvis, and appendicular bones, metaphysic long bone defects causing "Erlenmeyer flask deformity," "bone in bone" appearance of the vertebrae giving rise to "sandwich" vertebrae and "rugger-jersey" spine. In the dental setting, patients with osteopetrosis present with complications such as dental caries, premature tooth loss, delayed eruption of teeth, enamel hypoplasia, root malformations, and thickened lamina dura. Approximately two thirds of patients with autosomal-dominant type osteopetrosis (64%) had stomatological manifestations.[7] The risk of osteomyelitis increases in the presence of local infection such as odontogenic infection. Therefore dental treatment for patients with osteopetrosis, especially extraction of the teeth, must be carried out with great caution. In our case, manifestation of the disease begun with stomatological complaints, including multiple periods of exacerbations and remissions of osteomyelitis over a period of 7 months after teeth extraction.

The diagnosis of autosomal dominant osteopetrosis OAD is often made following a complication or following a radiological workup.[8]

Examination of the oral cavity in patients with benign osteopetrosis objectively results in delayed eruption and impaction of teeth, malformed non-eruptive teeth, and early tooth loss.

Dental extractions may be necessary due to poor oral hygiene and the above-mentioned dental abnormalities. However, their completion is difficult and usually followed by extensive bone loss leading to prolonged osteomyelitis and fistulas. Even erupting teeth can lead to serious infection such as orbital cellulitis.

When considering differential diagnoses, conditions such as metaphyseal dysplasia, pycnodysostosis, diaphyseal sclerosis, osteopathia striata, osteopoikilosis, melorheostosis, Camurati–Engelmann disease, and infantile cortical sclerosis should be evaluated. Each of these disorders can be distinguished through a combination of historical, clinical, and radiological assessments. In this particular case, other sclerosing disorders were excluded due to the absence of characteristic features such as dwarfism (associated with pycnodysostosis), cognitive impairment (linked to Camurati–Engelmann disease), and the specific presentation of long-bone osteopetrosis observed in Pyle diseas.

Only supportive care is available for benign osteopetrosis. Therefore, symptom management of benign osteopetrosis is important to increase patient survival.[7]

Conclusion

Osteomyelitis is a recognized complication of osteopetrosis. As previously mentioned, maxillary osteomyelitis is a rare occurrence. Should it present, evaluation for underlying conditions, particularly osteopetrosis, must be undertaken. Despite advancements in medical care leading to increased life expectancy, the rising incidence of this condition necessitates greater attention.

Since osteopetrosis cannot be prevented, dental considerations are paramount in mitigating common complications such as osteomyelitis. Radiological diagnosis is essential for confirming its presence, and dentists must exercise caution when planning extractions and surgeries. Preventive measures, including maintaining good oral hygiene, employing caries-preventive strategies, restorative treatments, and the application of topical fluoride, should be recommended. Extraction and surgical procedures should be avoided if possible to prevent complications such as osteomyelitis

and fractures. If absolutely necessary, these procedures must be performed under strict antibiotic coverage.

Limitations

This Case studies typically focus on a single patient, which may not represent thelarger population.

Findings from a single case may not be applicable to other cases or broader contexts.

Informed Consent was taken from the patient for using the facial images and radiographs

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Ethical clearance: As it is a Case Report ethical clearance is not required.

Conflict of Interest : None

References

1. Stark Z, Savarirayan R. Osteopetrosis. Orphanet journal of rare diseases. 2009 Dec;4:1-2.
2. Liu YP, Lin XH, Yan MY, Lin BQ, Zhuo MY. Debridement in chronic osteomyelitis with benign osteopetrosis: A case report. Experimental and Therapeutic Medicine. 2016 Nov 1;12(5):2811-4.
3. Sallies M, Titinchi F, Morkel JA. Osteopetrosis complicated by osteomyelitis of the maxilla: a rare case report and review of the literature.
4. Jayachandran S, Preethi M. Clinical spectrum of osteopetrosis with secondary osteomyelitis of the mandible: Report of two cases. Journal of Indian Academy of Oral Medicine and Radiology. 2018 Apr 1;30(2):165-8.
5. Celakil T, Dogan M, Rohlig BG, Evlioglu G, Keskin H. Oral rehabilitation of an osteopetrosis patient with osteomyelitis. Case Reports in Dentistry. 2016;2016(1):6930567.
6. Patil K, Mahima VG, Raina A, Mutneja P. Osteopetrosis: A case report. Int J Med Dent Case Rep. 2016;2:1-3.
7. OB B. Type II autosomal dominant osteopetrosis (Albers-Schoñberg disease): clinical and radiological manifestations in 42 patients. Bone. 2000;26(1):87-93.
8. Raiteb M, Elmrini S, Slimani F. Surgical management of chronic osteomyelitis with benign osteopetrosis: A case report. Annals of Medicine and Surgery. 2021 May 1;65.