CASE REPORT

Intermediate Type of Osteopetrosis in a Ten Year Old Boy

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Abstract

Osteopetrosis is a disease characterized by failure of osteoclasts of bone as a consequence bone modeling and remodeling is impaired, characteristically resulting in skeletal fragility despite of increased density, which also cause hematopoietic insufficiency, disturbed tooth eruption, nerve disturbances, syndrome and growth impairment. Human OP is a heterogeneous genetic disorder encompassing different molecular lesions and range of clinical features, which share single pathogenic nexus in the osteoclasts. We present a rare case of osteopetrosis in a 10-year-old boy with a palatal bony and infraorbital inflammatory swellings. The characteristic clinical, radiologic, and laboratory evaluation confirmed the diagnosis of intermediate osteopetrosis.

Keywords: Osteopetrosis, radiographic, jaw bones, palate.

INTRODUCTION

Osteopetrosis (OP) also known as Albers-Schonberg disease and marble bone disease, refers to group of genetic bone disorders with varying spectrum of clinical and radiological manifestations characterized by an increase in the density of all bones resulting in failure of primary spongiosa bone resorption and bone remodelling.[1] Although the precise nature of the disease is not known, several genes have been implicated in the etiopathogenesis of OP in humans.[1] The basic defect in all types of OP is the failure of osteoclasts to resorb bone at different phases. This lack of resorption results in the nonfunctional bone being grossly altered—the cortices are thickened, individual bony trabeculae are increased, marrow spaces are encroached upon, leading to paucity of hematopoietic tissue and consequent secondary anemia.[2] Thus the resulting bone is compromized with increased propensity to develop fracture and osteomyelitis. Osteopetrosis comprises a clinically and genetically heterogeneous group of conditions that share the hallmark of increased bone density on radiographs. OP is sub-classified into mainly three types.[2,3]

1. Severe infantile or malignant (autosomal recessive) type.
2. Benign or adult (autosomal dominant) type.
3. Intermediate (autosomal recessive) type.

The incidence of OP ranges from 1;200000 to 1;500000. Features like generalized retarded growth, hydrocephaly and prominent frontal bossing and unusual facial features in OP result because of increased bone density affecting the growth and development. Several oral manifestations of OP reported are delayed eruption, hypodontia, enamel hypoplasia, malformed roots, dental caries, defects in periodontal ligament, thickened lamina dura and osteomyelitis of jaws.[4] The radiographic features of the skull, chest and extremities in OP exhibit osteosclerosis of the entire skeletal system, including the base and widening of diploe of skull and endosteal bone formation in pelvis and scapula.[4,5] Over all the characteristic laboratory findings of various types include, altered or normal serum calcium and serum phosphorus levels but increased parathyroid hormone and acid phosphatase levels.[1-4,6-9] The diagnosis of OP is labelled considering the characteristic clinical, radiographic and laboratory findings, with adjunct use of genetic evaluation.[1] The prognosis of OP patients depends upon the type of disease. The current management modalities of OP include vitamin-D, gamma-interferon, erythropoietin, cortico-steroids and surgical modes like bone marrow trans-plantation with varying outcome.[1]

CASE REPORT

A 10-year-old boy along with his parents attended dental OPD of our college seeking treatment for swelling of the palate of 4 months and left infraorbital region of 3 days duration (Figs 1 and 2). The palatal swelling was gradual in onset, slowly progressive and painless interfering with speech and swallowing recently. The swelling gradually increased to attain the final size. The other swelling in the

left infraorbital region of 3 days duration was sudden in onset, progressive and associated with throbbing and continuous type of pain. Patient gave history of absence of many teeth since childhood and poor vision that was progressive from last 5 years. The family history revealed the consanguineous marriage of his parents as well as the death of his 4 years old elder sister because of hospitalization for high fever. She also had progressive blindness and total absence of teeth with unusual facial features. The other medical and personal histories were non-contributory.

On general physical examination patient was moderately built and poorly nourished, weighed 29 kg and 119 cm tall with hydrocephaly, prominent frontal bossing and bitemporal constriction. The ophthalmic findings included nystagmus, mild proptosis of the left eye along with continuous watering from the left eye. Moderate hepatosplenomegaly was also noted. Extraorally swelling was diffuse, firm and tender on the left side of the face below the infraorbital margin. Introrally the expanile bony hard palatal swelling was 3 × 3 cm occupying almost entire palatal surface with a deep cleft in between (Fig. 2). In the 62 region the mucosa showed surface ulceration attributed to unknown trauma. Dental examination revealed hypodontia with the presence of only seven teeth, which were hypoplastic, mobile and displaced maxillary teeth (62 and 53) due to palatal swelling. All the laboratory values of serum calcium (9.7 mg %), serum phosphorous (5.3 mg/dl) and parathyroid hormone (78.30 pg/ml) were marginally increased except serum acid phosphatase (25 IU/l), which was very high and hemoglobin was 9 gm%.

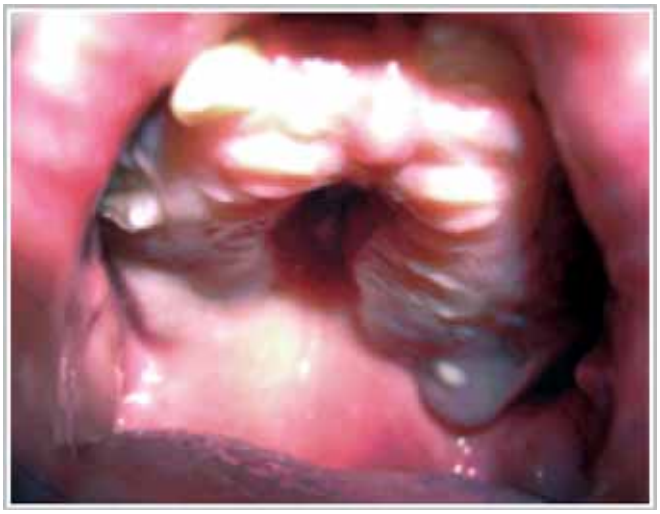

Fig. 2: Palatal swelling with deep cleft swelling

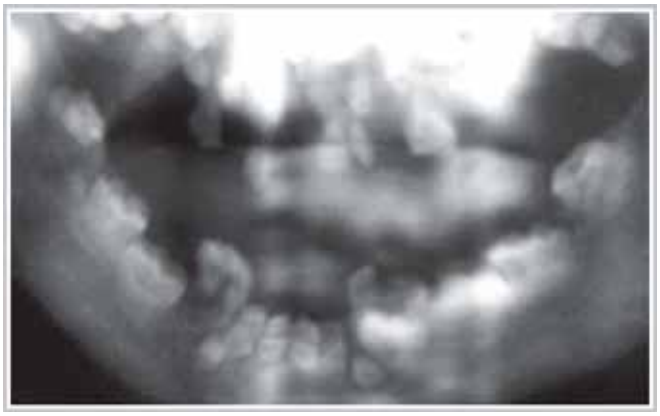

Fig. 3: OPG showing multiple impacted teeth with prominent osteosclerotic changes

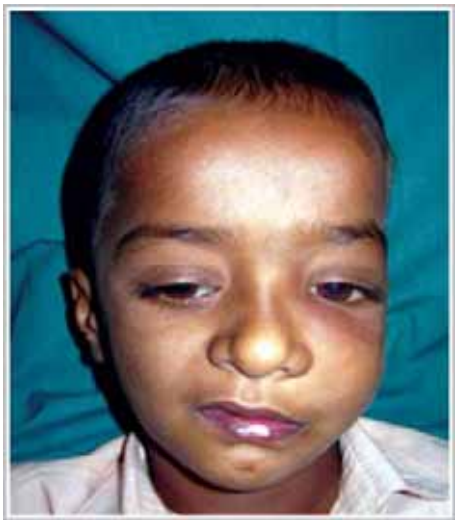

Fig. 1: Left diffuse infraorbital swelling

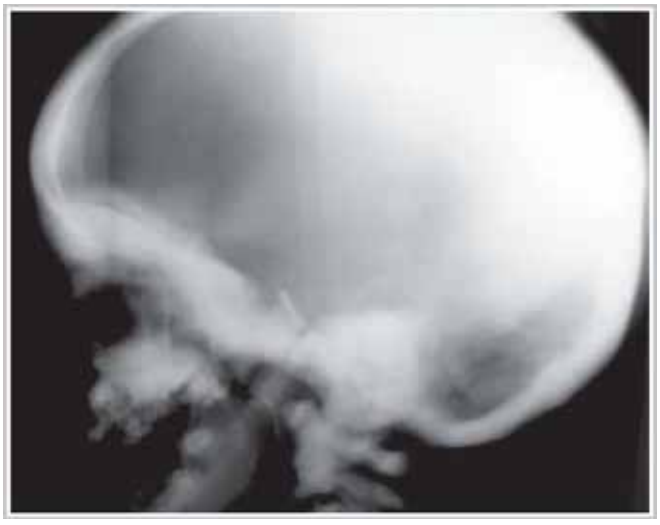

Fig. 4: Lateral skull showing sclerosis of base and calvarium

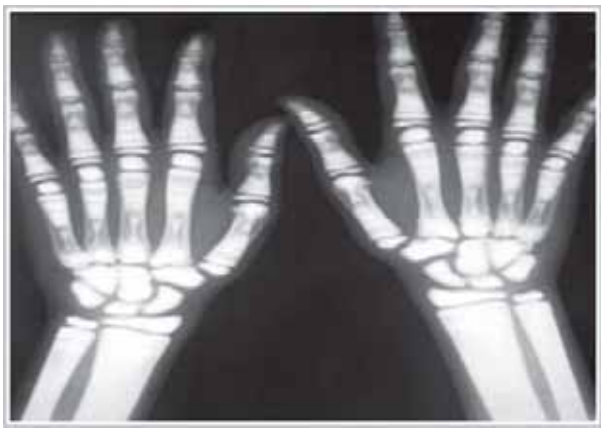

Fig. 5: Hand and wrist showing typical bone-in-bone and transverse bands

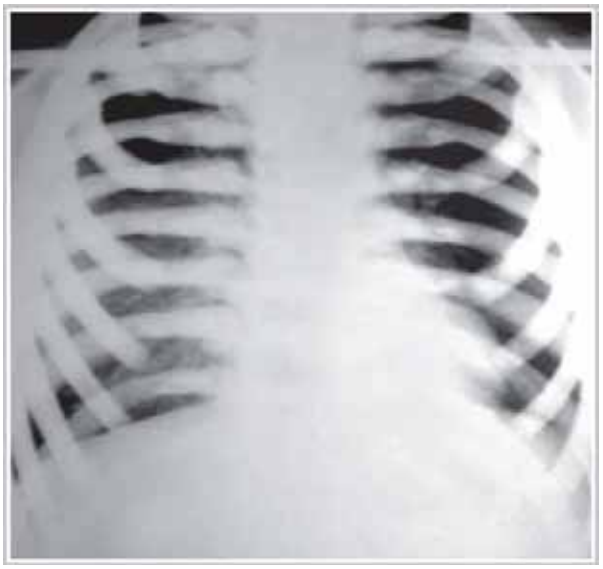

Fig. 6: Chest radiograph showing diffuse osteosclerosis

The radiographic features of OPG and lateral skull of the patient (Figs 3 and 4) revealed increased overall density of maxilla, base and vault of the cranium. The mandible showed diminished trabeculae and presence of multiple unerupted, hypoplastic teeth with malformed roots in both the arches. The chest radiographs revealed the overall increased density of ribs, spine and scapula. The classical 'bone in bone' or 'endobone' (rugger gersy sign) was seen in hand-wrist and transverse bands observed in pelvic radiographs (Figs 5 and 6). The final diagnosis of intermediate OP was done considering the overall clinical, radiographic and laboratory findings.With pediatrician consultation, antimicrobial drugs augumentin (amoxicillin + clauvanic acid) and metronidazole (metrogyl) IV and oral duoflam (ibuprofen and paracetamol) were given for the infraorbital swelling for 7 days which reduced considerably. Patient was under follow-up and he was advised for oral rehabilitaion, but parents of patient expressed inability for further treatment due to financial constraints.

DISCUSSION

Intermediate OP patients present with abnormal stature and tend to exhibit manifestations in end of first decade. Some of OP patients show cranial nerve deficits, macrocephaly, mild or moderate or severe anemia and abnormal teeth that may predispose to develop osteomyelitis of jaws.

Occasionally localized bony swellings in both the jaws are reported to occur in OP as found in this case may be due to the disease process[3,5] The characteristic widespread osteosclerosis of OP may also be seen in other disorders like Van Buchem disease, autosomal dominant osteosclerosis, pyknodysostosis, craniometaphyseal dysplasia and osteopathia striata, in this context the presence of BB-CK (brain isoenzyme creatine kinase) a molecular marker of OP may help to differentiate from others[1,8,9] The malformed teeth within sclerotic bone have potential to develop osteomyelitis of the regional bone. Early recognition and treatment of initial gingival, periodontal or soft tissue infection would prevent them developing into serious condition like osteomyelitis.

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