

CASE REPORT



Diagnosis behind the swelling of face in a 10-year-old child: A case report

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Abstract

Osteopetrosis is a rare genetic disorder of bone, characterized by impaired bone remodeling. Hyperostosis occurs due to lack of resorption of immature bone by osteoclasts. Abnormal remodeling of primary, woven bone to lamellar bone results in a brittle bone. In most ethnic groups, osteopetrosis occurs where consanguinity is common. The purpose of this article is to present a case of osteopetrosis and comprehensive review of its classification, clinical description, diagnosis, and management.

Keywords: Autosomal recessive, osteomyelitis, osteopetrosis

Introduction

A German radiologist, Albers-Schonberg first described osteopetrosis in 1904. It is a symptom complex related to altered bone metabolism. Different types are: An autosomal recessive malignant form, infantile malignant form, and intermediate osteopetrosis. Transient infantile osteopetrosis, carbonic anhydrase type II deficiency, lethal post infectious, and acquired are rare.

Features are fragile bones despite increase in bone mass, anemia, disturbance in tooth eruption, and impairment of growth.

The incidence of osteopetrosis is thought to be 1 in 100000 to 1 in 500000.^[1] Osteopetrosis shows a vast spectrum of clinical, physiologic, and genotypic expressions.^[2]

Case Report

A 10-year-old girl presented to the Department of Pedodontics with Preventive Dentistry with the chief complaint of painful swelling on the left side of face since 1 week [Figure 1].

There was a history of self-inflicted injury 1 week back. The child also had a history of delayed milestones of development. There were no significant findings among her parents or

siblings. The girl was born by a full-term normal delivery to a consanguineously married couple and elder sibling aged 13 years was healthy.

On general examination, the patient was found to be of short stature, poorly built, and nourished. There was increased head circumference, frontal bossing, depressed nasal bridge, distended abdomen, hypertelorism, and pigeon-shaped chest.

The patient had never gone to a dentist. A soft, diffuse swelling of approximately 2.5 cm × 2.5 cm was found on the left side of middle part of the face, which was tender on palpation. Pus discharge was evident immediately below the lower palpebral fissure of the left eye. Left submandibular lymph nodes were palpable and tender.

Intraoral examination revealed pale oral mucosa and few mobile deciduous teeth and none of the permanent teeth were visible. Clinically, primary maxillary lateral incisors, canines and left primary first and second molars and right primary first molar were present [Figure 2]. In the mandible, the primary canines, left and right primary first and second molars were present [Figure 3].

Laboratory investigations revealed normocytic hypochromic anemia. Abdominal ultrasonography revealed massive splenomegaly.



Figure 1: Clinical photograph of osteopetrosis patient showing swelling of face on left side and pus discharge below the left eye



Figure 3: Intraoral photograph showing mandibular primary canines, left and right primary first and second molars



Figure 2: Intraoral photograph showing primary maxillary lateral incisors, canines and left primary first and second molars and right primary first molar

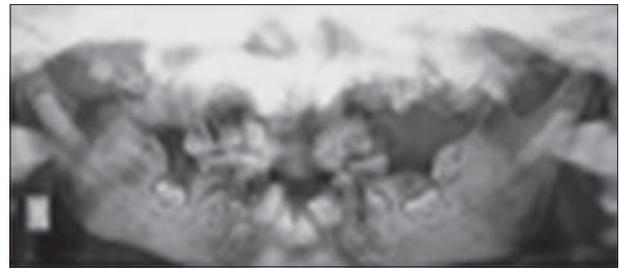


Figure 4: Orthopantomogram showing diffuse radiopacity in maxilla and morphologically abnormal multiple unerupted permanent tooth buds in maxilla and mandible

The orthopantomogram showed uniformly dense bone with an increased gonial angle and no differentiation between cortical and medullary bone. Maxilla and mandible showed morphologically abnormal multiple permanent tooth buds [Figure 4].

Radiographs of the skull [Figure 5], chest, hand-wrist anteroposterior view [Figure 6] and pelvis showed increased density of the entire skeleton with typical “bone within bone” appearance. Erlenmeyer flask deformity at distal end of long bones was a characteristic finding.

Based on the clinical, laboratory, and radiographic findings, it was diagnosed as autosomal recessive osteopetrosis. She was put on systemic antibiotic therapy, and local debridement of the affected jaw was done. The swelling was completely subsided after 5 days of medication and soft diet.

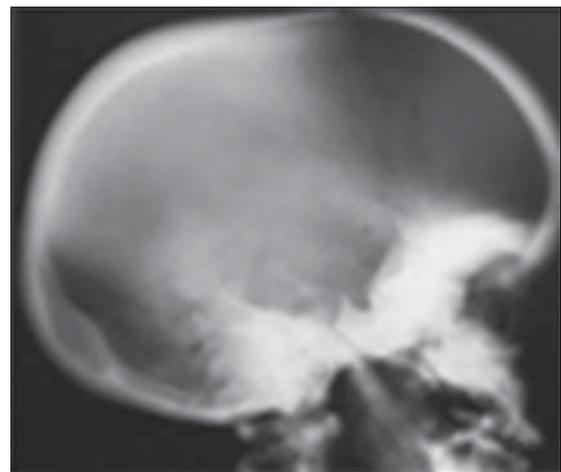


Figure 5: Radiograph of the skull showing increased density

Discussion

Osteopetrosis is a rare hereditary condition characterized by dense bone due to defective bone resorption. Malignant



Figure 6: Increased density hand-wrist anteroposterior view radiograph

recessive osteopetrosis is an uncommon disorder with incidence of 1:200000 population.^[3]

Key features are bone fractures, failure to gain normal visual milestones and hepatosplenomegaly. Other features include recurrent infections and abnormal craniofacial appearance.^[4] Impaired osteoclastic function may be due to a number of heterogeneous molecular or genetic defects.

Our patient was brought with signs of swelling of face on the left side and pus discharge below the left eye.

Bone defects and neuropathies related to cranial nerve entrapment occur due to failure of the skull foramina to widen completely. Nasal stuffiness is often the presenting feature. Manifestations include proptosis, deafness and hydrocephalus, and delayed dentition.^[5]

Hematological findings are due to obliteration of bone marrow cavity by bone causing anemia.^[5] In our case, hemoglobin

level was 5.8 g. Massive splenomegaly with coarse echogenecity was noted.

Radiologically, bone within bone appearance differentiates this disease from other sclerosing dysplasias.^[5] Osteomyelitis secondary to odontogenic infection is a common complication in infantile malignant osteopetrosis due to abnormal blood circulation.^[1]

Infants with malignant osteopetrosis also suffer from recurrent infections as a result of defect in macrophage function.^[2]

Conclusion

An autosomal recessive osteopetrosis is an uncommon hereditary bone disease, effective therapy must be individualized. Counseling of patients and parents on appropriate lifestyle modifications is needed to prevent fractures. Provide genetic counseling to parents to allow appropriate family planning.

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