ABSTRACT

Osteopetrosis is a rare genetic disorder that causes generalized sclerosis of bone due to a defect in bone resorption and remodeling. Osteomyelitis is well documented as a complication of osteopetrosis. This disease can be severe and difficult to treat in the osteopetrotic patient. Prevalence is about 0.005% in general population. There are relatively few cases reported of osteopetrosis and its actual cause is not known. Dental abnormalities that present radiographically are delayed eruption and missing teeth. The overall clinical and radiographic findings of this patient were sufficient to arrive at the diagnosis of osteopetrosis.

Keywords: Albers-Schonberg disease, Marble bone disease, Generalized sclerosis, Osteosclerosis.

INTRODUCTION

The term osteopetrosis is derived from the Greek ‘osteos’ meaning bone and ‘petros’ meaning stone. Osteopetrosis is an inherited, rare autosomal bone disorder of unknown etiology. It was first described in 1904 by German radiologist Albers Schonberg,1 hence the disease also gets the name Albers-Schonberg disease. This disorder includes impaired osteoclast function and marked increase in bone density. The incidence of osteopetrosis is thought to be 1 in 100,000 to 1 in 500,000. Osteopetrosis exhibits a vast spectrum of clinical, physiologic and genotypic expressions.2,3 Dental abnormalities that present radiographically are delayed eruption and missing teeth.4 The severe infantile forms of osteopetrosis are associated with diminished life expectancy, with most untreated children dying in the first decade as a complication of bone marrow suppression. The main complications are confined to the skeleton, including fracture and osteomyelitis with dental abscess or caries.5 This disease can be severe and difficult to treat. Treatment may include disfiguring procedures involving surgical removal of affected facial and skeletal bones. Radiologic features are usually diagnostic.6 The purpose of this paper is to report the case that presented to us in the 1st decade, involving maxilla and associated with odontogenic infection.

CASE REPORT

The patient was an 8-year-old boy who reported with the complaint of toothache and swelling with pus discharge from since 1 week. A detailed history was taken; patient had fractures of long bones twice after a fall, but had healed uneventfully. Family history revealed that the parents had consanguinous marriage. On examination there was an intraoral palatal swelling and pus discharge in relation to 64, 65 (Fig. 1). Many teeth were malformed and few were missing. A diagnosis of periapical infection was made. Radiographic investigation revealed diffuse sclerosis of bones. An orthopantomograph (OPG) confirmed the presence of generalized opacity in both the jaws. Lateral skull showed sclerosis of the base and cranial vault (Fig. 2), long bones radiograph showed dense radiopacity (Fig. 3), chest radiograph (Fig. 4) and pelvic region (Fig. 5) also showed diffuse generalized opacity. Hemoglobin was 8 gm%. Serum biochemistry values were in normal limit. Correlating the radiographic features with the clinical features, the case was diagnosed as osteopetrosis. Infection was controlled by administration of intravenous antibiotics and 64, which was grade III mobile was extracted.

DISCUSSION

Osteopetrosis is a genetic disorder that is divided into primary basic types: An autosomal dominant benign form and autosomal malignant recessive form. Rubin designated the malignant type as osteopetrosis congenita and benign type as osteopetrosis tarda.7 Another variant of osteopetrosis tarda was described and diagnosed in children or adults as the intermediate with poor prognosis2,6 which may manifest like the infantile type or be asymptomatic and seen on radiological survey as diffuse osteosclerosis.7 It is an
observed fact that, patients with rare systemic ailments like osteopetrosis rarely visit dentist unless they present with complications or secondary changes in oral and paraoral hard and soft tissues. This is very well illustrated in the present case where palatal swelling prompted the patients to seek treatment from dentist. Characteristic dental changes in osteopetrosis have been reported by many authors like the poorly formed and hypoplastic teeth, short roots, missing and unerupted teeth, early exfoliation of teeth, early onset periodontal and pulpal infections. Radiographic features of jaw anomalies, overall sclerotic appearance of jaws, most frequently maxilla, unerupted teeth, missing teeth, thickened cortices in calvaria, increased bone density of the cranial base, thickened trabeculae, thickening of the lamina dura diminished marrow spaces in our case coincide with findings reported by various authors. Patients with the disease seem to be susceptible to caries. The presented case findings and features are consistent with the intermediate type of osteopetrosis. There is no curative therapy for osteopetrosis though supplements of calcium, interferons, bone marrow transplants, have been tried but all with little success. Biopsy as a diagnostic procedure is not advisable because of the risk of developing osteomyelitis, hence no biopsy was performed.

Differential diagnoses of osteopetrosis are as follows:
- Craniometaphyseal dysplasia
- Pyknodysostosis
- Hypoparathyroidism
- Osteoblastic metastases
- Sclerosteosis
- Diaphyseal dysplasia
- Osteopathia striata

**SUMMARY**

The presented case may thus more accurately represent examples of intermediate osteopetrosis. The general dental practitioner or clinicians should be aware of such unusual presentation of osteopetrosis, hence thorough systemic, radiographic and laboratory evaluation of such systemic diseases is vital in improving the overall health of the patients.
REFERENCES


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