CASE REPORT

Osteopetrosis: A case report
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Abstract
Osteopetrosis is an uncommon skeletal disorder characterized by generalized sclerosis of bones due to defective osteoclast function. A wide variation in clinical severity of the disease may be noted. Radiographic features are usually diagnostic. A case of benign autosomal dominant form of osteopetrosis in an asymptomatic 14-year-old boy is hereby reported.

Keywords: Albers-Schonberg disease, endobone, marble-bone disease, osteopetrosis, Rugger-Jersey spine

Introduction
Osteopetrosis is a rare genetic skeletal disorder distinguished by a marked increase in bone density caused by a defect in bone remodeling due to failure of normal osteoclast function.[1] The benign autosomal dominant (tarda) form is frequently asymptomatic and diagnosed incidentally in late childhood or adulthood.[2,3] Radiography is an essential tool for diagnosis of osteopetrosis, in which the bones appear uniformly dense and sclerotic.[4] We hereby report an interesting case of benign autosomal dominant type of osteopetrosis in a 14-year-old boy.

Case Report
A 14-year-old boy reported to us for a routine dental evaluation. Medical history revealed that the patient was an epileptic and on phenytoin tablets (100 mg OD) since a year. Patient’s surgical and family history was insignificant.

Examination disclosed a left cephalic asymmetry [Figure 1a] with no other significant oro-dental findings. A routine radiographic evaluation comprised of an orthopantomograph and skull views to assess the cephalic irregularity. Panoramic radiograph [Figure 1b] showed diffuse sclerosis of the maxillary and mandibular alveolar bones. An anteroposterior and lateral skull views depicted an increase in density of cranial and base of the skull bone [Figures 1c and 2a]. In view of these findings, the patient was subjected to a complete skeletal survey that constituted radiographs of the chest, pelvis, limbs and dorsolumbar spine. The bones showed a uniformly increased radiodensity with the lack of clear distinction between cortical and cancellous bone [Figures 2b and 3a and 3b]. Increased sclerosis was noted at the upper and lower borders of the vertebrae giving a pathognomonic Rugger-Jersey spine appearance [Figure 4]. Considering these characteristic radiographic findings in an asymptomatic 14-year-old, a diagnosis of osteoporosis – tarda variety was rendered.

Furthermore, a thorough pediatric evaluation of the patient was unrevealing. A complete hemogram divulged no abnormalities. Biochemical analysis disclosed normal serum calcium and phosphorus levels. Marked elevation of alkaline phosphatase (1, 177.6 IU/L) documented to occur in osteopetrosis was noted.

Since the patient was asymptomatic, no therapeutic interventions were made. The patient has been educated about the nature of the disease and its probable complications. He is currently under close follow-up.

Discussion
Osteopetrosis eponymously known as Albers-Schonberg disease, marble bone disease, and osteopetrosis generalisata
was first described by a German radiologist, Albers-Schonberg, in 1904.\textsuperscript{[1,5,6]} Osteopetrosis is a rare genetic disorder resulting in generalized sclerosis of bones with a reduction of marrow spaces subsequent to defective osteoclast function and decreased bone turnover.\textsuperscript{[6]} The ensuing change in bone structure is accompanied by a striking tendency toward fragility with fractures following trivial trauma.\textsuperscript{[6]}

Osteopetrosis is categorized chiefly into three types: Infantile malignant autosomal recessive form, intermediate autosomal recessive form, and the benign autosomal dominant form or tarda.\textsuperscript{[3,4,6]} The first two types are associated with a poor prognosis, have an onset at or shortly after birth and are fatal.\textsuperscript{[6,8]} On the contrary, the benign autosomal dominant form manifests in later childhood and is incidentally discovered on a radiographic examination as seen in the case hereby reported.\textsuperscript{[1,6,8]}

Approximately, 40% of the afflicted are asymptomatic as in the case presented.\textsuperscript{[1]} Others may present with recurrent fractures associated with minor trauma, bone pain, and osteomyelitis often affecting the mandible.\textsuperscript{[7]} Failure of remodeling of the skull bones causes narrowing of the skull foramina that may result in cranial nerve compression leading to optic nerve atrophy and blindness, deafness and facial paralysis.\textsuperscript{[1]}

Radiographic features include increase in bone density of the entire skeleton (inclusive of the jaws, skull base, ribs, pelvis, clavicle, femur, and humerus) with defective metaphyseal remodeling.\textsuperscript{[2,6]} Long bones exhibit decreased marrow spaces with cortical thickening and become club-shaped.\textsuperscript{[6]}

Within bone” or “endobone” appearance reflecting the dense shadow of the abnormal new bone within the outline of the normal bone is archetypal of osteopetrosis.\textsuperscript{[2,6]} Radiographically, autosomal dominant osteopetrosis is categorized into two subtypes – Types I and II.\textsuperscript{[8]} Massive sclerosis of the skull and thickening of the cranial vault characterize Type I while Type II is distinguished by sclerosis of the skull, most prominent at the base. In Type II, the spine shows thick dense endplates at the upper and lower borders of the vertebrae with a normal appearance in the middle third referred to as a “Rugger-Jersey spine.”\textsuperscript{[8]} Based on the radiographic presentation of the disease, our patient was diagnosed as having sporadic autosomal dominant Type II osteopetrosis. Delayed eruption of teeth, congenitally missing teeth, poor tooth calcification, hypercementosis, and premature loss of teeth constitute the dental changes in osteopetrosis.\textsuperscript{[7]} Increased thickness of the lamina dura and an increased tendency for tooth decay has also been reported.\textsuperscript{[7]} Osteomyelitis is a noticeable complication of osteopetrosis owing to impaired vascularity of bone.\textsuperscript{[6]} In an osteopetrotic, osteomyelitis is commonly associated with dental extractions or surgical exposure of the pathologic bone most often the mandible.\textsuperscript{[6,7]}

In contrast to the infantile forms, myelophthisic anemia caused by bone marrow insufficiency and hepatosplenomegaly resulting
from extramedullary hematopoiesis are rare in the tarda group of patients. [1,4] Accordingly, our patient had all hematological values within normal limits. Elevated serum alkaline phosphatase, low serum phosphorus levels, and hypocalcemia have been reported in osteopetrosis. [2] Hypocalcemia may also be the cause for seizures occasionally. [5] Serum calcium and phosphorus levels were normal in our case. The alkaline phosphatase levels were markedly elevated in our case probably due to contributing factors of phenytoin therapy and adolescent age of the patient in addition to his osteopetrotic state. Chronic phenytoin therapy may induce osteoblast proliferation leading to skull thickening in 34% of patients with seizure disorders. [5] However, since our patient was on phenytoin therapy only since a year the possibility of the same causing increased skull thickness was marginally low.

Treatment of the benign form of osteopetrosis is purely symptomatic, intended to correct facial deformity or recurrent fractures. [4] Neuropathy related to nerve compression may be surgically treated. [4] In the event of an osteomyelitic process, therapy with high-dose antibiotics accompanied with thorough debridement of the necrotic tissues and hyperbaric oxygen has been advocated. [6] Bone marrow transplantation is the only definitive treatment for patients with the infantile malignant form. [2,4] Gamma interferon, low calcium diet, oral cellulose phosphate and calcitriol may also benefit this group of patients. [2,4]

**Conclusion**

Osteopetrosis is an uncommon skeletal disorder. Although radiographs are diagnostic, a thorough history and clinical evaluation of the patients is essential. Rare asymptomatic forms as in the case reported may be diagnosed incidentally. An early diagnosis aided by relevant investigations may help prevent the occurrence of future complications in this group of patients.

**References**