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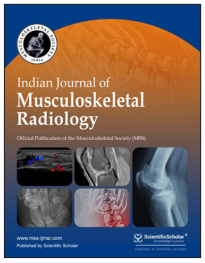

Case Report

Magnetic Resonance Imaging Diagnosis of Osteopetrosis in a Child Presenting with Blindness

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ABSTRACT

Osteopetrosis is a rare hereditary bone dysplasia of heterogeneous pathophysiology in which failure of osteoclastic bone resorption leads to increased bone mass, which has poor mechanical properties. Patients present in childhood with complaints of bone pains, failure to thrive and growth retardation. Other clinical findings include severe anemia, hepatosplenomegaly, lymphadenopathy and thrombocytopenia. The dense, extremely brittle bones fracture easily. Involvement of the cranium can lead to optic nerve atrophy with blindness or other cranial nerve defects. The diagnosis is primarily radiographic and may be supported by computed tomography scan. We are reporting a case of a 4-year-old boy who presented with blindness since the age of 6 months and the diagnosis of osteopetrosis was suggested on magnetic resonance imaging.

Keywords: Osteopetrosis, Magnetic resonance imaging, Blindness

INTRODUCTION

Osteopetrosis is a rare hereditary and familial bone abnormality characterized by the lack of resorption of normal primitive osteochondrous tissue. The persistence of this tissue inhibits the formation of normal mature adult bone with a medullary canal containing marrow tissue.[1] Sclerotic brittle bones are formed as a result. Anemia is invariably present and does not correlate well with the degree of sclerosis.

The diagnosis remains radiographic and can be supported by computed tomography.[2]

CASE REPORT

A 4-year-old boy presented to the ophthalmologist with the complaints of blindness since the age of 6 months. There was also a history of failure to thrive. He was the firstborn child and did not have any sibling. There was no such history in the first-order relatives. On fundoscopic examination, bilateral discs appeared pale with attenuation of vessels around it, suggestive of optic atrophy. The patient was then referred to us for magnetic resonance imaging (MRI) of the brain and orbits. On MRI evaluation of head, thickened sclerotic calvarium was noted [Figure 1]. Bilateral optic nerve canals were being encroached on by the thickened calvarium [Figure 2]. Mastoid air cells were non-pneumatized with fluid in the middle ear cavities [Figure 1]. Cerebral parenchyma, ventricles, and subarachnoid spaces were normal. Further, the patient's skeleton was evaluated on X-ray. The skull in anteroposterior and lateral views showed thickening and sclerosis of the skull base [Figure 3]. X-ray of the lower extremities revealed increased density of

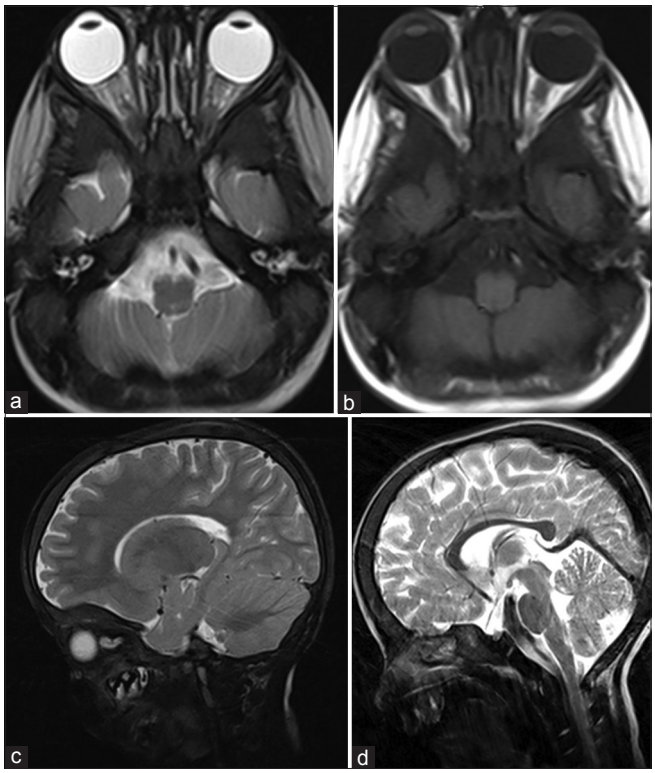

Figure 1: Turbo spin echo (TSE) T2W (a) and SE T1W (b) images show thickened sclerotic calvarium. Mastoid air cells are non-pneumatized. Fluid is seen in the middle ear cavities. TSE T2W images in parasagittal (c) and sagittal (d) planes demonstrate the thickened bone encroaching on the paranasal sinuses, small pituitary fossa, and unerupted teeth and also note the hypointense cervical vertebrae.

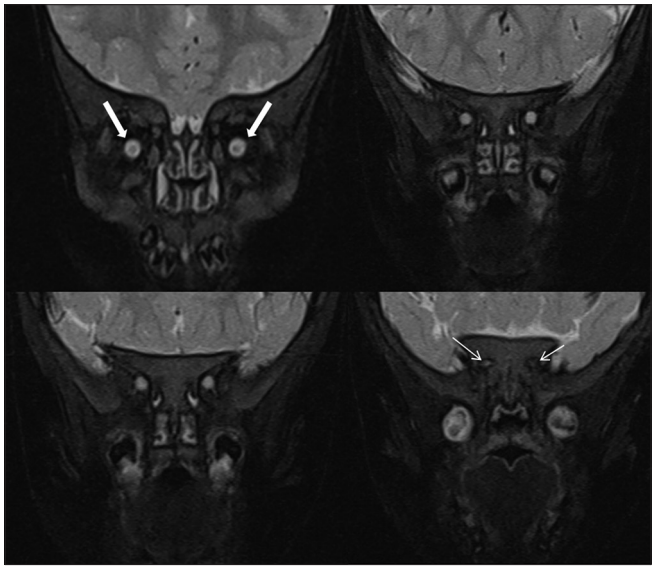

Figure 2: High-resolution FS turbo spin echo T2W images in coronal plane show marked atrophy of optic nerves (thick arrows) with severe stenosis of the optic nerve canals bilaterally (thin arrows).

the bones, bone within a bone appearance, and Erlenmeyer flask deformity of the femora [Figure 4]. X-ray AP view of the pelvis with bilateral hip joints revealed increased bone density and a bone within a bone appearance of iliac bones, the femoral heads, and supra-acetabular regions [Figure 5]. Lateral view of the dorsolumbar spine revealed dense bands along the superior and inferior endplates [Figure 6].

DISCUSSION

Osteopetrosis is more commonly known as marble bone disease. It results from more than one genetic or biochemical defect, with at least five types of the disease having been described.[3]

The two most commonly seen forms are autosomal recessive (malignant) osteopetrosis (AROP) and autosomal dominant (benign) osteopetrosis (ADOP).

AROP is associated with deficient osteoclastic resorption of the primary spongiosa. This form of osteopetrosis usually

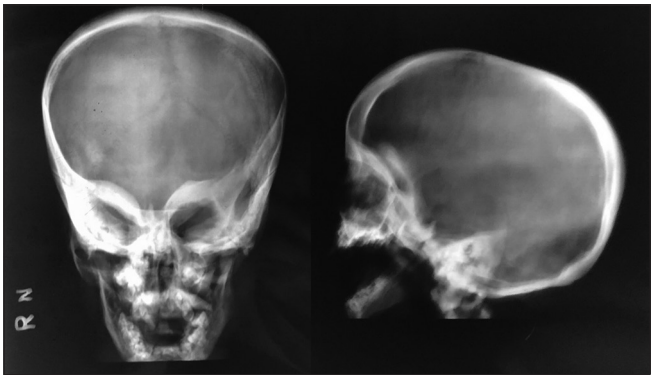

Figure 3: Anteroposterior and lateral radiographs of skull confirm thickening and sclerosis of the skull base.

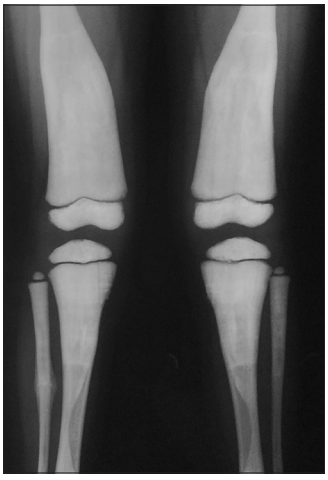

Figure 4: X-ray of the lower extremities revealing increased density of the bones, bone-within-a-bone appearance, and Erlenmeyer flask deformity of the femora.

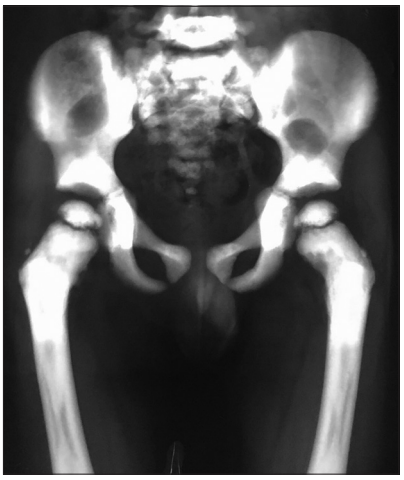

Figure 5: X-ray of the pelvis reveals increased bone density and a bone within a bone appearance of iliac bones, the femoral heads, and supra-acetabular regions.

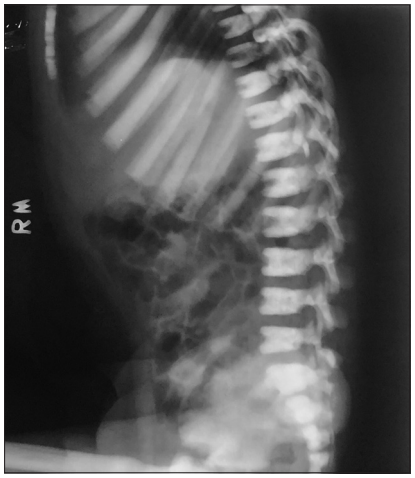

Figure 6: X-ray of dorsolumbar spine in lateral view shows dense bands along the superior and inferior endplates (sandwich vertebrae).

begins in infancy. It is also known as infantile or malignant osteopetrosis. Patients exhibit poor osseous growth and remodeling with anemia, infection, and hemorrhage resulting due to obliteration of the marrow spaces.[4] Bones are fragile and fracture easily. Dentition may be delayed. Other manifestations of AROP are optic atrophy, cranial nerve palsies, stenosis of the petrous internal carotid artery, the jugular vein within narrowed jugular foramina, and the cervical vertebral arteries within narrowed transverse foramina.[5] Cranial nerve entrapment neuropathies occur due to failure of the foramina in the skull to widen completely. Manifestations include deafness, proptosis, blindness, and hydrocephalus. Sleep apnea and blindness due to retinal degeneration are also seen. Radiological features usually are diagnostic.

A more uncommon form of the disease, intermediate osteopetrosis (IOP), also demonstrates autosomal recessive inheritance. However, this variant tends to present later in childhood than the more malignant form. Typically, these patients have short stature and experience more malignant features of malignant osteopetrosis.[4] Frequently, IOP is distinguished from the malignant form only when a milder clinical course evolves with age.

ADOP is also known as Albers-Schönberg disease. It typically has onset in late childhood or adolescence.[6] The adult type may be asymptomatic and discovered incidentally on plain radiographs when fractures occur. Few patients with ADOP experience more severe complications of the disease, including facial nerve palsy, visual loss, deafness, bone pain, and osteomyelitis. Two types of ADOP have been classified on the basis of clinical and radiographic criteria. Type I ADOP is characterized by diffused calvarial sclerosis and thickening with diffuse sclerosis of bone in the spine and pelvis. Histologically, few abnormalities are apparent in the modeling of trabecular bone. Indeed, the bones are quite strong and fractures are rare. In type II ADOP, calvaria is less severely involved, but the skull base is sclerotic and thickened.[7] It classically displays the radiographic sign of dense bands of sclerosis parallel to the vertebral endplates also known as "sandwich vertebrae."[6]

Other radiographic features include a "bone-within-a-bone appearance," resulting from a reduced ability of the osteoclasts to model and remodel the bone. Bone-within-a-bone appearance may also be seen in Paget disease, sickle cell disease, thalassemia, lead poisoning, acromegaly, congenital syphilis, and hypervitaminosis D.[8]

The expanded diametaphyseal areas of femora are homogenously sclerotic also known as "Erlenmeyer flask deformity." Erlenmeyer flask deformity has been reported in a heterogeneous group of craniotubular bone dysplasias, as well as commonly seen in other systemic disorders including Gaucher disease, Niemann-Pick disease, and thalassemia, and has been reported as a late change in lead poisoning.[9]

Radiotracers have also been utilized to assess osteopetrosis. 99mTc-sulfur colloid scintigraphy is used to show bone marrow distribution of the disease. 99mTc-MDP is used to show uptake at fracture sites and splayed metaphyses of long bones.[10]

The only cure for osteopetrosis is bone marrow transplant. Patients must receive a bone marrow transplant early enough in life such that complications of osteopetrosis can be reversed or prevented altogether. Bone marrow transplant in infantile osteopetrosis may be followed by reversal of optic canal stenosis and preservation of vision.

CONCLUSION

Osteopetrosis is a rare hereditary bone dysplasia commonly diagnosed on radiography. Our endeavor through this case report is to highlight the role of MRI in suggesting the diagnosis of osteopetrosis if it is the first modality, a patient has been referred for.

Acknowledgments

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Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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