An unusual cause of optic atrophy in a child

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A 13-year-old child presenting with gross visual impairment was diagnosed as a case of optic atrophy. However, radiological investigations revealed osteopetrosis, which, though rare, can result in optic atrophy. The aim of this case report is to highlight this possibility while evaluating cases of optic atrophy in young patients.

Key words: Optic atrophy, osteopetrosis

Osteopetrosis can cause optic nerve compression in the bony optic canal and lead to optic atrophy. Timely awareness about the condition could possibly guide the clinician to measures related to prevention of optic atrophy.

Case Report

A 13-year-old male child presented at this tertiary care center with gradual onset loss of vision over a period of 3 years. There was no history of pain, redness, watering, floaters, photopsia, ocular or head trauma, prior ocular surgery, systemic illness/chronic drug therapy, headache, vomiting, or seizures. There was history of frequent fractures in the past with minimal trauma (right clavicle and right arm). There was no history of similar ocular/visual problems in the family. However, there was history of multiple fractures with trivial trauma in elder brother. On examination, the child was fairly built and moderately nourished. Pectus excavatum was present. There was no hepatosplenomegaly. He was evaluated for hearing loss by ENT specialist and found normal. His vision in RE was 20/120 improving to 20/60 and in LE counting finger at 4 feet improving to 20/200 with RAPD in LE. On fundus examination, optic discs were normal in size, circular, and pale with distinct margin. On confrontation test, there was constriction of fields in both eyes. Colour vision (Ishihara chart) and contrast sensitivity (Pelli Robson's chart) were reduced in both eyes. VER (flash) in RE showed 4 μV (amplitude), 114 ms (latency), and in LE was 2 μV, 144 ms. Pattern VER in RE showed 3 μV, 134 ms, while in LE there was an extinguished response. His hematological parameters were normal. Based on the above findings, a clinical diagnosis of primary optic atrophy was made keeping a differential diagnosis of compressive optic neuropathy, hereditary optic atrophy, toxic, and nutritional optic neuropathy.

Axial [Fig. 1a] and coronal [Fig. 1b] computed tomography (CT) scan brain images showed an abnormally increased density of all the skull bones with narrowing of bilateral optic canal (white arrows). Skeletal survey done subsequently also showed diffusely increased bone density of all bones, confirming the diagnosis of osteopetrosis. Diameter of bony optic canal was 3.8 mm in the right side and 3.5 mm on the left side. Normal average transverse diameter of the optic canal is 3.57 ± 0.61 mm and the longitudinal diameter is 4.82 ± 0.38 mm.[1] T2W sagittal MR [Fig. 2] better demonstrated compression of optic nerve sheath complex on both sides (white arrows). There was no sign of raised intracranial pressure or hydrocephalus. In background of recurrent fractures and optic canal narrowing with abnormal increased density of bones, the diagnosis of compressive optic neuropathy secondary to osteopetrosis-induced optic canal narrowing was made.

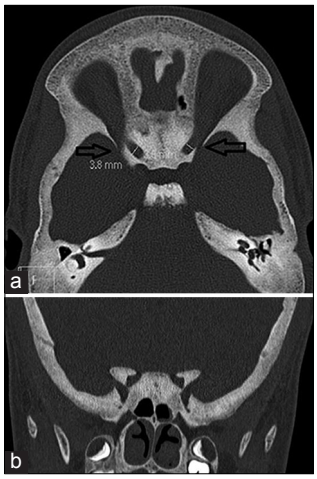

Figure 1a and b: Axial and coronal CT scan brain images show an abnormally increased density of all the skull bones with narrowing of bilateral optic canal (white arrows)

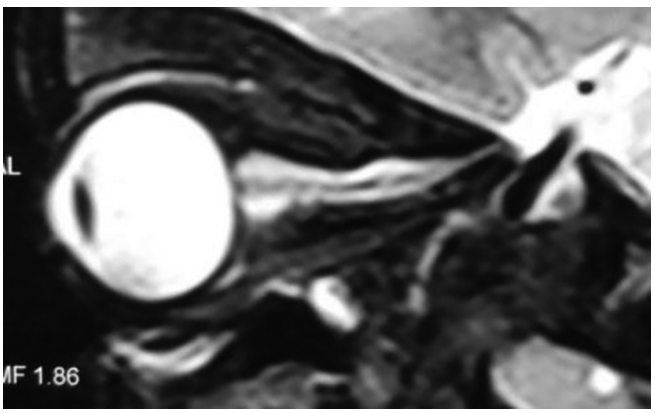

Figure 2: T2W sagittal MRshowing compression of optic nerve sheath complex on both sides (white arrows).

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Discussion

Osteopetrosisis was caused by reduced activity of osteoclasts which results in defective remodelling of bone and increased bone density.[2] The defect in bone turnover results in skeletal fragility despite increased bone mass, and it may also cause hematopoietic insufficiency, disturbed tooth eruption, nerve entrapment syndromes, and growth impairment. Sclerosis of bones and narrowing of bony foramina leads to compressive neuropathies.

Typically osteopetrosis presents with fractures**,** short stature, and hypocalcemia, pancytopenia and optic nerve compression and rarely hypogenitalism.[3] There are three clinical groups of osteopetrosis, namely, infantile-malignant autosomal recessive, intermediate autosomal recessive, and autosomal dominant type. The incidence of autosomal recessive osteopetrosis is 1 in 250,000 births and autosomal dominant osteopetrosis is 1 in 20,000 births.[4] Investigations recommended are CT scan (brain, orbit), MRI (brain, orbit), radionuclide bone scan, genetic analysis, iliac crest bone biopsy, and skeletal survey. Patient was treated with high-dose calcitriol to stimulate osteoclast differentiation and anoptic canal decompression was planned. Optic canal decompression in the early stage of visual deterioration may reverse vision loss. Bone marrow transplant has been reported as a successful treatment option to widen the narrow optic canal in infantile osteopetrosis.[5]

It has been emphasized that optic nerve decompression should be wide and include not only unroofing of the bony canal but also drilling along both sides of the optic nerve as well as smoothening of the thick, irregular, and highly domed orbital roof by high-speed drilling to facilitate surgical exposure with minimal retraction of the frontal lobe.[6]

The aim of this case report is to highlight the possibility of optic nerve compression due to optic canal narrowing in cases of osteopetrosis. Optic canal decompression may be considered as a prophylactic measure to preserve vision in partial optic nerve atrophy or in fellow eye.

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