Bilateral endoscopic optic nerve decompression in an infant with osteopetrosis

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Osteopetrosis is a rare disorder of bone remodeling characterized by defective resorption leading to excessive skeletal mass including optic canal. Compression of the optic nerve from the narrowed optic canal is the most common cause of vision loss in children with osteopetrosis. We report the case of a 6-month old girl with osteopetrosis who underwent bilateral optic canal decompression via endoscopic transcaruncular approach for progressive deterioration of visual function secondary to compressive optic neuropathy from narrowed optic canals. The patient showed improvement in visual function postoperatively.

A 6-month old girl with Williams syndrome, congenital heart disease and hypercalcemia, and autosomal recessive infantile osteopetrosis (TCIRF1, c.304del.G, c.1887+G.A) was referred to the Children's Hospital of Pittsburgh Department of Ophthalmology for evaluation. Since her first ophthalmology examination at 2 months of age, she had developed roving nystagmus and variable strabismus. She had no response to light stimulus in either eye. Anterior segment examination was unremarkable. Fundus examination showed mild pallor of optic disk in the right eye with grayish dysplastic looking optic nerve in the left eye. Retinae were hypopigmented, with normal macula and vasculature. Flash visual-evoked potentials (VEP) revealed decreased amplitude in both eyes with prolonged latencies, worse in the left than right eye. Flash electroretinogram (ERG) was normal bilaterally. Computed tomography (CT) revealed thickening of the optic strut and narrowing of both optic canals, to about 3 mm in diameter (Figure 1). Magnetic resonance imaging (MRI) showed bilateral optic nerve hypoplasia, with compression of optic nerves from thickened optic canals, thinning of the corpus callosum, dysplastic cerebellum and frontal horns, and a type 1 Chiari malformation. In the presence of severe narrowing of both optic canals, with evidence of decreasing visual function, our team offered bilateral endoscopic optic canal decompression (OCD). She underwent bilateral OCD via endoscopic transcaruncular approach. The right optic canal was decompressed first, after performing right medial orbital decompression through transcaruncular incision; decompression was carried out anterior to the face of the chiasm and along the contralateral optic canal. Three months postoperatively visual acuity had improved to 20/2700 bilaterally by Teller acuity cards. She was fixating with the left eye, but there was improvement in nystagmus and visual responses bilaterally. The right eye had restrictive strabismus. At final follow-up, 7 months after the decompression, she continued to maintain the visual acuity of 20/2700 bilaterally. Repeat CT scan showed open optic canals with no bony regrowth (Figure 2). Postoperative flash VEP also showed improvement in latency and amplitude, mostly in the left eye (Figure 3).

Discussion

Infantile osteopetrosis is a genetic disorder with benign dominant and malignant recessive forms.[1] The main defect in osteopetrosis is poor osteoclastic activity with abnormal bone resorption, which causes generalized increase in skeletal density, including the skull base. Vision loss in the most common presenting symptom, seen in 75% of children with osteopetrosis.[1] The etiology of visual impairment is multifactorial but due primarily to optic atrophy associated with narrowing of the optic canals.[2-5] Papilledema, primary retinal degeneration, and vascular compression have also been postulated as possible causes of visual impairment in children.[6] The etiology of optic atrophy in patients with osteopetrosis is controversial, as is surgical intervention.

The diagnosis of progressive visual loss in preverbal children is difficult. In infants with osteopetrosis it mainly depends on the assessment of visual the child's behavior, change in the appearance of optic nerve, narrowing of the optic canal on imaging, and, most importantly, on visual-evoked responses.

At 6 months of age, our patient showed a change in visual behavior, with roving nystagmus and poor response to light. Negative ERG ruled out retinal degeneration as a cause of visual impairment. MRI of the brain did not reveal hydrocephalous or other signs of raised intracranial pressure except Chiari I malformation. Increasingly abnormal VEP and clearly narrowed optic canals prompted OCD.

In their series of 15 children with osteopetrosis, Thompson and colleagues[5] found that the most frequent early indication of visual impairment was delay in VEP latency, which often precedes the fundus changes of optic neuropathy and optic nerve changes on neuroimaging. Of 15 patients, only 1 had OCD after 2 unsuccessful bone

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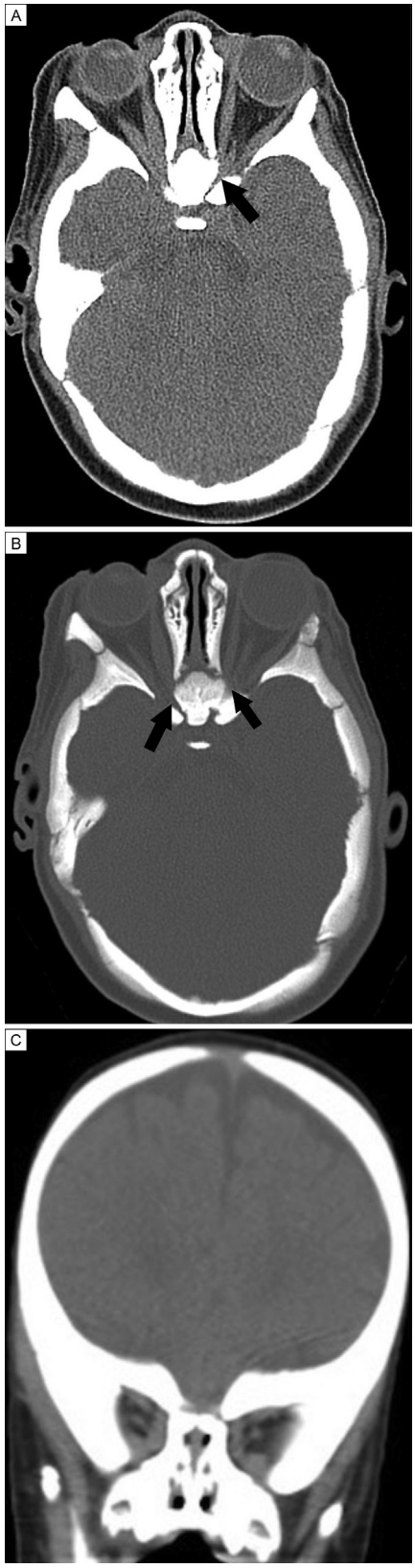

FIG 1. Preoperative computed tomography (CT) scans. A, Axial cut, soft tissue window; B, Axial cut, bone window, showing thickening of orbital and skull bones and narrow optic canals bilaterally (black arrows). C, Coronal cut showing narrowing of the posterior orbit due to thickened bones.

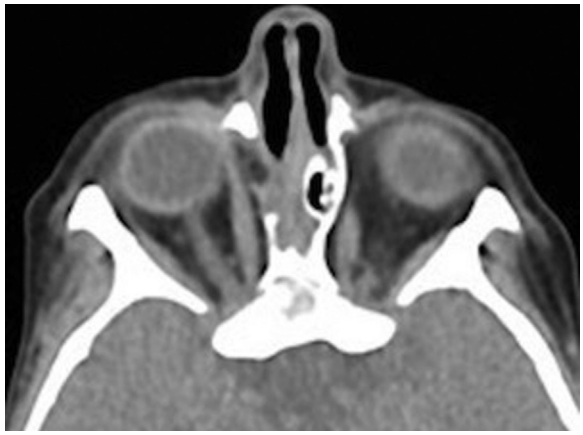

FIG 2. Postoperative CT scan showing patent optic canals bilaterally and decompressed right medial orbital wall.

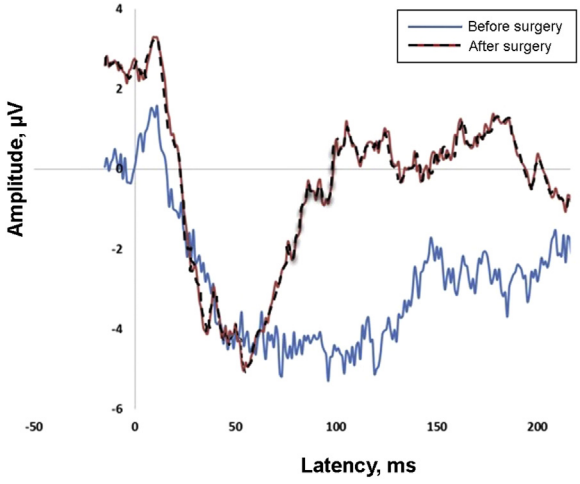

FIG 3. Flash visual evoked potential of the left eye showing improvement in amplitude and latency postoperatively.

marrow transplantations before 6 months of age who died from a severe vascular complication at the time of surgery.[5]

The indications for OCD in children are relatively rare and include compression due to extrinsic tumors (eg, sphenoid wing meningioma), traumatic optic neuropathy, bony lesions (eg, fibrous dysplasia), and osteopetrosis.[7] Recently the minimally invasive endoscopic endonasal approach has gained popularity over conventional anterior skull base surgery via frontal or frontolateral craniotomy. This technique provides direct access to both optic canals without manipulation of the optic nerve and avoids an external scar.[8] In our patient, with a small nose, the transcaruncular approach provided a wider working corridor.

OCD with good visual outcomes has been reported more frequently in older children with osteopetrosis.[2,3,6] It has rarely been performed in infants, with only 5 patients <12 months of age reported before the advent of endoscopic surgeries for skull base lesions.[1,2,4,5] Of the 5

Table 1. Infants with osteopetrosis who underwent optic canal decompression reported in literature

Case	Age, months	Sex	Indication for surgery	Visual acuity		VEP		Follow-up	Ref. No
				Pre-op	Post-op	Pre-op	Post-op		
1	6	F	OA + OC narrowing	NA	NA (stable)	NA	NA	Died at 15 mos	1
2	4	M	OA + OC narrowing	NA	NA	Abnormal	Improved	Died	4
3	7	F	OA + OC narrowing	NA	NA	Abnormal	NI	Died at 2 mos	4
4	8	M	OA + OC narrowing	NA	Normal	Abnormal	Improved	4 months	4
5	1	M	OA + OC narrowing after 2 BMTs	NA	NA	Abnormal	NI	Died at 6 mos	5

BMT, bone marrow transplantation; *NA*, not available; *OA*, optic atrophy; *OC*, optic canal; *VA*, visual acuity; *VEP*, visual-evoked potential.

patients, 4 showed improvement in VEP; however, there was no documentation of quantitative assessment of visual acuity (Table 1). To our knowledge, ours is the first case of OCD via endoscopic approach in an infant with osteopetrosis.

Prophylactic deroofing of the optic canal has been reported in other conditions associated with excess bony growth of the optic canal; however, the procedure carries a risk of vision loss.[9] The general consensus is to intervene surgically only in patients with sudden or progressive deterioration of vision.[10] A recent metanalysis comparing surgery and observation in patients with fibrous dysplasia showed that expectant management resulted in better visual outcomes than OCD in asymptomatic patients with radiological evidence of optic nerve compression.[10]

Although visual impairment in our patient was multifactorial, she had postoperative improvement in behavioral indicators of visual function as witnessed by the parents and vision teachers as well as improved VEP, visual acuity, and nystagmus. In centers with an experienced team of skull base surgeons, endoscopic OCD should be considered in children with optic neuropathy secondary to osteopetrosis. Regular ophthalmology evaluation is crucial to recognize early visual dysfunction in these children. The importance of electrodiagnostic testing and neuroimaging cannot be overemphasized if the child shows any change in visual behavior.

Literature Search

PubMed was searched on November 30, 2017, for English-language results, using the following terms: *osteopetrosis*, *optic atrophy*, and *optic canal decompression*.

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Fresh frozen plasma (Octaplas) and topical heparin in the management of ligneous conjunctivitis

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Ligneous conjunctivitis is a rare form of chronic recurrent membranous conjunctivitis with reduced plasminogen activity. It is

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