CASE REPORT

Visual Function Improvement After Optic Nerve Sheath Fenestration in Osteopetrosis Patients with Optic Canal Stenosis: A Report of Two Cases

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ABSTRACT

The outcome of two patients with visual loss from osteopetrosis who underwent an optic nerve sheath fenestration (ONSF) is reported. A 20-year-old male and 26-year-old female with osteopetrosis had optic nerve edema. Computed tomography and magnetic resonance imaging demonstrated optic canals stenosis. Both patients underwent unilateral ONSF. After ONSF, the patients experienced improvement in visual acuity and optic nerve appearance. Therefore, when papilledema is recognized in osteopetrosis patients, it may be reasonable to start with an ONSF even if the optic canal seems to be stenotic because of the lower morbidity that is associated with this procedure compared with other surgical options.

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Introduction

Osteopetrosis is a heterogeneous bone disorder characterized by osteoclast dysfunction and impaired bone resorption that cause increased bone density.^{1–3} The disorder leads to bone marrow dysfunction and life-threatening conditions such as anaemia, pancytopenia, and sepsis.^{3,4} Other disabling features such as blindness and deafness occur as well, mainly due to nerve compression in the bony canals. Optic nerve atrophy can be a consequence of compressive neuropathy, long-standing papilledema caused by increased intracranial pressure, or due to retinal degeneration.^{3,5,6}

The optimal management of visual deterioration has been a matter of controversy.^{6–8} Optic nerve sheath fenestration (ONSF) was suggested as a treatment option for papilledema when the optic canals are not stenotic.⁸ We describe two patients with visual deterioration from papilledema and significant optic canal narrowing that improved after ONSF.

Case report

Case one

A 20-year-old man with osteopetrosis presented to our ophthalmology clinic. He reported a gradual

decline in visual acuity in his right eye. He has been blind in this eye for around 1 month prior to his presentation. Additionally, he reported visual obscuration in his left eye. His right visual acuity was no light perception, and his left visual acuity was 20/20. The Ishihara colour vision test score was 0/15 for the right eye and 3/15 for the left eye. He had full extraocular motility with concomitant mild sensory exotropia. The anterior segment examination result was within normal limits bilaterally. Fundus examination revealed a pale disc in the right eye and grade 2 papilledema in the left eye (Figure 1A). Humphrey visual field perimetry showed an enlarged blind spot in the left eye. Magnetic resonance imaging (MRI) of the head showed generalized bone thickening, hyperdense facial and cranial bones, and markedly narrowed optic nerve foramina, more on the right side than on the left side, with larger optic nerve sheaths on the left side (Figures 2 and 3). The lumbar puncture opening pressure was 31 cm H₂O with normal cerebral spinal fluid analysis.

ONSF was performed on the left eye. Two months after the ONSF, reduction of the papilledema (Figure 1B), stable vision, and improvement of colour vision to 7/15 were observed, without any reported visual obscuration. The patient's vision continued to be 20/20 after 4 years of follow-up.

Figure 1. (A) Photographs of optic nerves at presentation, Case 1. The right eye shows a pale atrophic disc. The left eye shows an optic disc edema and tortuous retinal blood vessels. (B) The right and left optic discs after optic nerve sheath fenestration OS. The left optic disc shows a reduction of the optic nerve edema.

Figure 2. Axial T2-weighted image shows dilated optic nerve sheath bilaterally more obvious on left side.

Case two

A 26-year-old woman with osteopetrosis was referred to our ophthalmology clinic because of reduced vision. She noticed blurred vision in her left eye 2 months before her appointment. On examination, her visual acuity was 20/25 in the right eye, corrected with a pinhole to 20/20, and

counting fingers at 5 ft in the left eye, corrected with a pinhole to 20/100. The Ishihara colour vision test score was 15/15 in the right eye and 4/15 in the left eye. She had an afferent pupillary defect in her left eye. Motility and intraocular pressure were normal. Cornea examination results showed signs of keratoconus. Fundus examination revealed grade 1–2 papilledema bilaterally but worse in the left eye (Figure 4A). Humphrey visual field perimetry showed an enlarged blind spot in the right eye and an unreliable test result in the left eye. Computerized tomography scan and MRI revealed severe diffuse thickening of the skull base and vault. Severe narrowing of the skull base foramina and tonsillar herniation measuring 6 mm were observed. Bilateral narrowing of the internal auditory canals and optic canals, with subsequent compression and atrophy of the optic nerves, was also evident (Figures 5 and 6). Magnetic resonance venography revealed no signs of thrombosis. Lumbar puncture was not performed because of the risk

Figure 3. Axial T1 weighted image: Left image shows hyperostosis (right arrow) resulting in narrowing of the optic canals bilaterally, and thinning of the optic nerves (left arrow). Right image: Axial T1 weighted image of a normal person for comparison shows normal optic nerves and optic canals.

Figure 4. (A) Optic nerve photographs at presentation. The right and left sides show grade 1–2 optic nerve edema. (B) The right and left optic nerves 2 months after optic nerve sheath fenestration of the left eye. Both sides show reduction of the optic nerve edema. The left optic disc shows temporal pallor.

Figure 5. Coronal and axial T2-weighted images show dilated optic nerve sheaths (right arrow) bilaterally, and thinning of the optic nerves (left arrow).

Figure 6. Axial (left) and coronal (right) CT images show grossly hyperostotic skeletal structures with narrow optic canals bilaterally.

of herniation. The patient was referred to a cornea specialist for keratoconus management.

ONSF of the left eye was performed 2 weeks later. Post-ONSF, the visual acuity was 20/20 in the right eye and 20/60 in the left eye. The colour vision test score in the left eye improved to 8/15. A marked reduction of the bilateral disc swelling was observed (Figure 4B). Visual fields were also improved. Although increased intracranial pressure could not be confirmed by lumbar puncture, the subarachnoid fluid around the optic nerves on MRI and the positive response to ONSF support this assumption. Three months later, a hemi-cranioplasty was performed for headache. Her vision had continued to stabilize by her last visit 9 months later.

Discussion

Progressive visual loss is common in osteopetrosis. As the life expectancy of osteopetrosis patients is increasing owing to advances in the management of the disease, preservation of functional vision as long as possible is highly important. The role of regular ophthalmic evaluation in the prevention of visual dysfunction is imperative, as early detection and treatment might contribute to good recovery.

The visual deterioration in osteopetrosis is often due to compressive optic neuropathy caused by the bony overgrowth and narrowing of the optic canals and foramina. Several other mechanisms contribute to the visual loss, including long-standing papilledema, which leads to optic nerve atrophy. Therefore, determining the cause of the

progressive visual deterioration is crucial in the management of these patients.

Decompression of the optic canal with or without deroofing is commonly suggested for cases of optic canal narrowing, but it carries significant mortality and morbidity risks. On the other hand, patients with visual loss due to papilledema with patent optic canals may respond to medical treatment or surgical procedures such as ONSF. Successful ONSF was reported in a patient with papilledema where the optic canals were not significantly stenotic.⁸

To our knowledge, no previous cases of ONSF in patients with optic canal narrowing have been reported. Both of our cases are unique in that they had significant optic canal narrowing and papilledema as mechanisms responsible for the visual loss, with good response to ONSF. Our patients' vision improved after ONSF; thus, it is reasonable to conclude that increased intracranial pressure was a contributing factor to the visual deterioration. While direct optic nerve compression may indeed be responsible for the visual decline in our patients, papilledema in these two patients that resulted from increased intracranial pressure caused by either thickening of the skull or secondary to outflow obstruction was an exacerbating factor and, therefore, was promptly managed when identified. However, more cases and longer follow-up are needed to reveal the usefulness of ONSF in osteopetrosis patients with papilledema in the presence of narrowed optic canals.

In conclusion, when papilledema is recognized in osteopetrosis patients, initiation of ONSF may be reasonable even if the optic canal seems to be

stenotic because the morbidity associated with this procedure is lower than those associated with other surgical options.

Declaration of interest

The authors declare that there are no conflicts of interest. The authors alone are responsible for the writing and content of the article.

References

1. Tolar J, Teitelbaum SL, Orchard PJ. Osteopetrosis. *New England J Med* 2004;351(27):2839–2849. doi:[10.1056/NEJMra040952](https://doi.org/10.1056/NEJMra040952).
2. Felix R, Hofstetter W, Cecchini MG. Recent developments in the understanding of the pathophysiology of osteopetrosis. *Eur J Endocrinol* 1996;134(2):143–156. doi:[10.1530/eje.0.1340143](https://doi.org/10.1530/eje.0.1340143).
3. Del Fattore A, Cappariello A, Teti A. Genetics, pathogenesis and complications of osteopetrosis. *Bone* 2008;42(1):19–29. doi:[10.1016/j.bone.2007.08.029](https://doi.org/10.1016/j.bone.2007.08.029).
4. Van Wesenbeeck L, Van Hul W. Lessons from osteopetrotic mutations in animals: impact on our current understanding of osteoclast biology. *Crit Reviews™ Eukaryot Gene Expr* 2005;15(2):133–162.
5. Siatkowski RM, Vilar NF, Sternau L, Coin CG. Blindness from bad bones. *Surv Ophthalmol* 1999;43(6):487–490. doi:[10.1016/S0039-6257\(99\)00048-X](https://doi.org/10.1016/S0039-6257(99)00048-X).
6. Hoyt CS, Billson FA. Visual loss in osteopetrosis. *Am J Dis Child* 1979;133(9):955–958.
7. Hwang J-M, Kim I-O, Wang K-C. Complete visual recovery in osteopetrosis by early optic nerve decompression. *Pediatr Neurosurg* 2001;33(6):328–332. doi:[10.1159/000055980](https://doi.org/10.1159/000055980).
8. Allen RC, Nerad JA, Kattah JC, Lee AG. Resolution of optic nerve edema and improved visual function after optic nerve sheath fenestration in a patient with osteopetrosis. *Am J Ophthalmol* 2006;141(5):945–947. doi:[10.1016/j.ajo.2005.11.037](https://doi.org/10.1016/j.ajo.2005.11.037).