Bilateral Endoscopic Endonasal Optic Nerve Decompression in an Infant with Osteopetrosis: A Case Report

Yara Yammine, MD[1,2] ; Rita Maria Jalkh, MD[1,2]; Houssein Darwich, MD[1,2]; Zeina Korban, MD[1,2]
[1]American University of Beirut , [2]American University of Beirut Medical Center, Beirut, Lebanon

Abstract

***Background*:** Osteopetrosis is a rare genetic disorder causing abnormal bone density, often leading to vision loss due to optic canal stenosis and optic nerve compression; early intervention is crucial to prevent irreversible damage.

***Case Description*:** A 7-month-old male with osteopetrosis and optic nerve compression underwent bilateral expanded endoscopic endonasal optic canal decompression after radiological and genetic confirmation.

***Conclusion*:** Endoscopic endonasal optic nerve decompression is a safe, effective option for infants with osteopetrosis, offering minimal morbidity and significant potential for visual recovery, as demonstrated in the youngest reported case in the literature.

Introduction

Osteopetrosis is a rare inherited disorder, with the most severe cases typically autosomal recessive[1].
Osteopetrosis is characterized by increased bone density due to defective osteoclast function[2]. The failure of osteoclasts to resorb bone leads to the accumulation of dense but brittle bone, resulting in bone fragility, anemia, hepatosplenomegaly, and narrowing of cranial foramina and subsequent nerve entrapment[3].
Early diagnosis and intervention are crucial to prevent irreversible complications such as vision loss. Treatment can include medical management, hematopoietic stem cell transplant, and surgery to alleviate symptoms and prevent progression[4]. In cases where optic nerve compression is present, surgical decompression may be necessary and urgent to prevent permanent vision loss.

This case report highlights the successful management of optic nerve compression in an infant with osteopetrosis using bi-nostril endoscopic endonasal decompression, emphasizing the importance of early intervention and the effectiveness of this minimally invasive approach, especially in resource-limited settings.

Case Presentation

A 7-month-old male presented with hepatosplenomegaly, leukocytosis, thrombocytopenia, poor weight gain, and a family history of osteopetrosis with a known **CLCN7** mutation. Radiologic studies confirmed diffuse bony sclerosis, and genetic testing revealed a homozygous variant in **CLCN7**, consistent with autosomal recessive osteopetrosis.
By April 2024, MRI confirmed bilateral optic nerve compression and ophthalmologic exams showed progressive vision decline, including esotropia and pale optic nerves.
Given the disease's progression, the patient underwent bilateral expanded endoscopic endonasal optic canal decompression on July 25, 2024, successfully addressing the compression.
Due to the patient’s young age and small anatomical structures, the ophthalmology team had opted against the transcaruncular approach.

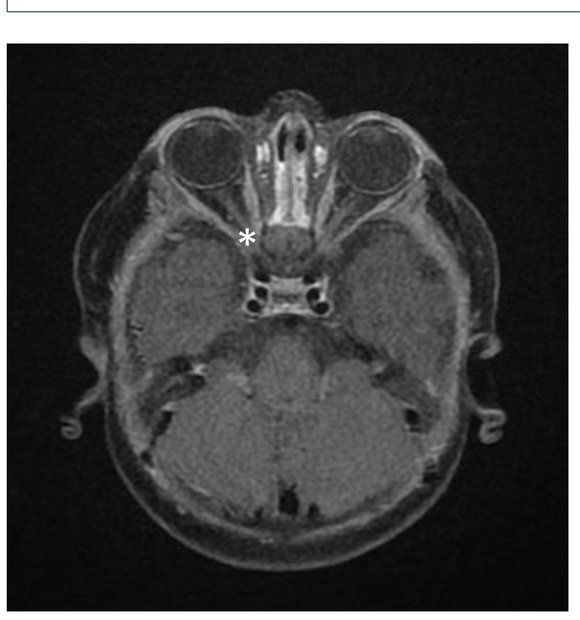

Figure 1. MRI Axial T1-Weighted: Narrowed Optic Canals

Intra- and Post-Operative Course

The surgical team, comprising rhinology, endoscopic skull base, and neurosurgery specialists, successfully performed bilateral expanded endoscopic endonasal optic canal decompression with no intraoperative complications, such as bleeding, CSF leaks, or optic nerve injury. Despite financial constraints preventing the use of navigation, the surgery was completed with precision. Under general anesthesia, a 30-degree, 2.7 mm endoscope was used to access the nasal cavities. A posterior septectomy was first performed. To improve exposure, the anterior one-fourth of the middle turbinate was resected bilaterally, with careful removal of ossified bone using a drill. Then, bilateral total ethmoidectomies were done. This allowed access to the medial orbital walls bilaterally, which were meticulously removed exposing the periorbita to decompress the orbits, extending the decompression posteriorly to the annulus of Zinn. Complete decompression of the optic canals was achieved bilaterally. Hemostasis was ensured using FloSeal, Surgicel, and Gelfoam.
Postoperative recovery was uneventful with no complications. Follow-up assessments revealed significant improvement in the patient’s vision, with enhanced visual tracking and responsiveness by August 5, 2024. The patient was cleared for planned bone marrow transplantation.

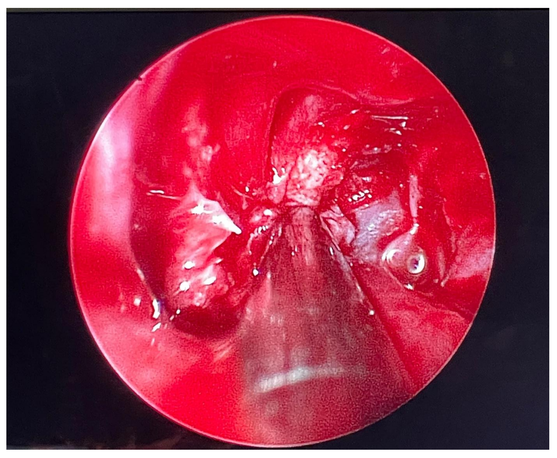

Figure 1. Nasal cavity view (bilateral medial orbital walls exposed)

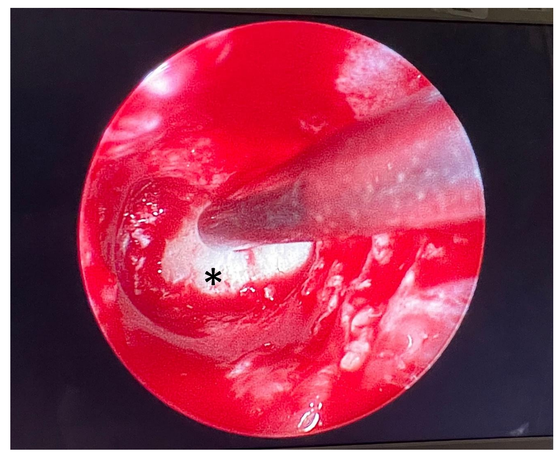

Figure 2. Left periorbita exposed posteriorly to annulus of Zinn

Discussion

Optic Nerve Decompression in Osteopetrosis
Various surgical approaches, including transcranial, transorbital, and transcaruncular methods, have been used for optic nerve decompression in osteopetrosis, each with unique risks and benefits. The expanded endoscopic endonasal approach offers a minimally invasive alternative with reduced recovery time and fewer complications[5], as demonstrated in this case.

Role of Navigation in Endoscopic Surgery
Navigation systems enhance precision in endoscopic surgery but are not always essential[6]. Experienced surgeons can achieve excellent outcomes without navigation by relying on detailed anatomical knowledge, making this approach more accessible in resource-limited settings.

Significance of Early Intervention
Early surgical intervention is crucial to preserving vision in young patients with optic nerve compression. Previous cases in children aged 1 to 5 years showed significant visual improvement with early decompression[7]. This case, involving a 7-month-old infant, highlights the importance of timely diagnosis and treatment to prevent irreversible damage, even in the youngest patients.

Cost Considerations
High costs of advanced procedures and limited resources in developing countries pose significant barriers. Affordable solutions, including international collaborations and training programs, can improve access to life-changing procedures like endoscopic optic nerve decompression in resource-limited settings.

Conclusion

This case report demonstrates the successful management of optic nerve compression in an infant with osteopetrosis, youngest reported in the literature, using a minimally invasive endoscopic endonasal approach, highlighting the importance of early intervention and surgical expertise in achieving excellent outcomes, even in resource-limited settings.

Contact

Zeina Korban MD
American University of Beirut
Riad El Solh 1107 2020, Beirut, Lebanon
zk27@aub.edu.lb
+961-1-350000 ext. 5471

References

1. Villa, A., Guerrini, M. M., Cassani, B., Pangrazio, A., & Sobacchi, C. (2009). Infantile malignant, autosomal recessive osteopetrosis: The rich and the poor. *Calcified Tissue International, 84*(1), 1-12.
2. Stark, Z., & Savarirayan, R. (2009). Osteopetrosis. *Orphanet Journal of Rare Diseases, 4*(1), 5.
3. Tolar, J., Teitelbaum, S. L., & Orchard, P. J. (2004). Osteopetrosis. *New England Journal of Medicine, 351*(27), 2839-2849.
4. Gerritsen, E. J., Vossen, J. M., Fasth, A., et al. (1994). Bone marrow transplantation for autosomal recessive osteopetrosis. *Bone Marrow Transplantation, 13*(4), 429-433.
5. Medsinge, A., Sylvester, C., Tyler-Kabara, E., & Stefko, S. T. (2019). Bilateral endoscopic optic nerve decompression in an infant with osteopetrosis. *Journal of AAPOS : The Official Publication of the American Association for Pediatric Ophthalmology and Strabismus, 23*(1), 40–42.
6. Shibata, T., Tanikawa, M., Sakata, T., & Mase, M. (2018). Urgent Optic Nerve Decompression via an Endoscopic Endonasal Transsphenoidal Approach for Craniopharyngioma in a 12-Month-Old Infant: A Case Report. *Pediatric neurosurgery, 53*(3), 182–187.
7. Hwang, J. M., Kim, I. O., & Wang, K. C. (2000). Complete visual recovery in osteopetrosis by early optic nerve decompression. *Pediatric Neurosurgery, 33*(6), 328–332.