CASE REPORT

Optic nerve tortuosity associated with posterior staphyloma in high myopia: a case report

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Abstract

We report on a case of bilateral tortuous optic nerve associated with posterior staphyloma and high myopia in a 50-year-old woman. Magnetic resonance imaging was used to evaluate potential etiologies. Posterior staphyloma is the most likely etiology of bilateral tortuous optic nerve.

Key words: Magnetic resonance imaging; Myopia, degenerative; Optic nerve

Case presentation

In October 2024, a 50-year-old Chinese woman was referred to Hong Kong Eye Hospital following an incidental finding of bilateral optic nerve kinking with partial empty sella on computed tomography of the brain to rule out idiopathic intracranial hypertension (IIH) after an episode of dizziness and syncope. The patient had a history of amblyopia and cataract in the left eye and high myopia (-13 D) in both eyes. She had undergone laser-assisted in situ keratomileusis in the left eye. The axial length was 29.6 mm in the right eye and 36.8 mm in the left eye. She was asymptomatic despite the bilateral optic nerve kinking. She had no headache, decline in visual acuity, or transient visual loss. Optic nerve function was preserved, with no relative afferent pupillary defect. Ishihara test revealed full and equal color vision in both eyes. She had no visual field defect. Fundus examination showed bilateral tilted discs with no disc swelling. The patient had no café-au-lait spots, neurofibromas, freckles, or any ocular features suggestive of neurofibromatosis type 1 (NF1). Her intraocular pressure was within the normal range.

Magnetic resonance imaging (MRI) of the brain and orbit showed bilateral tortuous optic nerves, with mild (<2 mm) cerebrospinal fluid filling of the optic nerve sheaths, partial empty sella, and bilateral posterior staphyloma (**Figure**). There were no other radiological features of IIH, and the subarachnoid space was unremarkable. No optic glioma was identified.

A lumbar puncture, performed by a neurosurgeon, showed a normal opening pressure of 19 cmH_2O, which is not indicative of IIH. Clinically, the patient had no ocular symptoms of IIH, as evidenced by the absence of headache, visual loss, and papilledema.

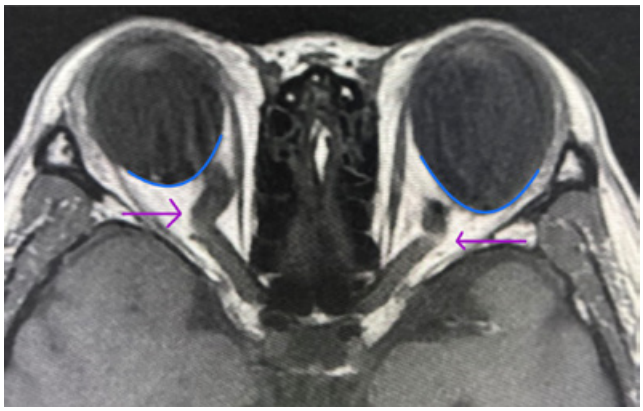

Figure. T1-weighted magnetic resonance imaging of the brain and orbit showing bilateral optic nerve tortuosity (arrows) and bilateral posterior staphyloma (curves), more prominent in the left eye.

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The bilateral optic nerve tortuosity was most likely the result of bilateral posterior staphyloma secondary to high myopia. The patient has been followed up at a neuro-ophthalmology clinic and has had stable visual acuity and no new complaints.

Discussion

In our patient, posterior staphyloma was the most likely etiology of the bilateral tortuous optic nerve, which is a benign and usually asymptomatic condition characterized by an abnormal curvature of the optic nerve, typically recognized on cross-sectional MRI.

Tortuous optic nerve is frequently reported in patients with NF1, a genetic disorder affecting the skin and central nervous system. Typical ocular features of NF1 include optic glioma and Lisch nodules, which are iris hamartomas and can be detected by slit lamp examination.[1] Patients with NF1 have an increased risk of tortuous optic nerve,[2] with incidences ranging from 12% to 31%.[3,4] Tortuous optic nerve in NF1 is characterized by a lack of congruity of the optic nerves in more than one coronal section and dilation of the subarachnoid space surrounding the optic nerves.[5] In our patient, the absence of cutaneous and ocular features of NF1 and unremarkable subarachnoid space on MRI strongly suggested that NF1 was not the cause of the bilateral tortuous optic nerve.

Another cause can be IIH, which is characterized by increased intracranial pressure without an identifiable cause such as a brain tumor or hydrocephalus. Classical presentations of IIH include headache, transient vision loss, papilledema, elevated opening pressure in a lumbar puncture, and the absence of a known cause of raised intracranial pressure.[6] Tortuous optic nerve in IIH is thought to result from increased cerebrospinal fluid pressure transmitted into the optic nerve sheath, causing distension and mechanical deformation of the nerve.[7] A meta-analysis identified tortuous optic nerve as a highly specific sign of IIH, with pooled specificity of 88.4%, although pooled sensitivity was only 36.9%.[8] Given the high specificity, tortuous optic nerve in our patient was strongly suggestive of IIH; however, the absence of headache, visual acuity deterioration, and papilledema in addition to the normal opening pressure in the lumbar puncture suggested against IIH.

Secondary intracranial hypertension due to tumors, cerebral venous sinus thrombosis, or other intracranial pathologies can give rise to tortuous optic nerve through mechanisms analogous to IIH.[9] Diagnosis of secondary intracranial hypertension depends on identification of the underlying cause of raised intracranial pressure. However, our patient had normal opening pressure in the lumbar puncture and no intracranial pathologies on multiple neuroimages.

Patients with glaucoma have a higher incidence of optic nerve kinking, particularly in those with normal tension glaucoma than in those without known optic nerve diseases (86% vs 18%).[10] Myopic glaucomatous optic neuropathy typically demonstrates glaucomatous changes of the optic nerve head, including neuroretinal rim thinning and retinal nerve fiber layer loss, accompanied by optic neuropathy manifestations such as progressive visual field defects. In contrast, in isolated tortuous optic nerve associated with high myopia, the optic disc appearance, optical coherence tomography parameters, and visual field testing results are generally within normal limits.

Other rarer causes of tortuous optic nerve include spaceflight-associated neuro-ocular syndrome, which is found in approximately 47% of astronauts after a long spaceflight.[11]

We hypothesize that our patient's bilateral tortuous optic nerve is due to bilateral posterior staphyloma, which is not a well-established etiology, and no study has shown their causative relationship. However, structural changes in posterior staphyloma may influence the configuration of the optic nerve. Staphylomas refer to abnormal protrusion of the eye's outer wall, typically involving thinning and bulging of the sclera or cornea, with underlying uveal tissue visible through the weakened area. Posterior staphyloma is an outpouching or localized ectasia of the sclera at the posterior pole of the eye, primarily associated with high myopia.[12] Scleral ectasia alters the normal anatomical contour of the globe and the optic nerve head, causing aberrant mechanical traction and possible optic nerve tortuosity or kinking.[13] Mechanistically, the staphyloma-induced scleral deformation displaces and stretches the optic nerve, resulting in a non-linear tortuous course as the nerve conforms to the altered globe shape.[13] Axial elongation in high myopia, the most common cause of posterior staphyloma, can lead to gaze-induced optic nerve head deformation and, potentially, tortuous optic nerve.[14]

Tortuous optic nerve is often benign and asymptomatic, with minimal functional impact on visual acuity, color vision, and visual fields. Nevertheless, it could be clinically significant if it is associated with NF1 or IIH. Patients with NF1 and tortuous optic nerve may be at higher risk of developing optic nerve glioma, although the association is not strongly supported by evidence.[15] Although tortuous optic nerve is a specific sign of IIH, the associated visual dysfunction is most likely due to elevated intracranial pressure and subsequent papilledema, rather than tortuous optic nerve itself.

This case report has several limitations. First, a single case precludes any definitive conclusion about causality between posterior staphyloma and tortuous optic nerve. The proposed association should be regarded as a hypothesis. Second, high myopia could be a confounding factor, as it is associated with multiple structural alterations of the posterior segment and optic nerve that mimic tortuosity. Third, neuroimaging cannot exclude subtle or coexisting etiologies of tortuous optic nerve, and the mechanical influence of posterior staphyloma on the optic nerve course remains inferential in the absence of longitudinal or biomechanical data. Finally, our findings may not be generalizable to other patients with

tortuous optic nerve or posterior staphyloma.

Contributors

All authors designed the study, acquired the data, analyzed the data, drafted the manuscript, and critically revised the manuscript for important intellectual content. All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

Conflicts of interest

As editors of the journal, CKMC and HKLY were not involved in the peer review process. Other authors have disclosed no conflicts of interest.

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Data availability

All data analyzed in this study are available from the corresponding author upon reasonable request.

Ethics approval

The patient was treated in accordance with the tenets of the Declaration of Helsinki. The patient provided written informed consent for all treatment and procedures and for publication.

References

1. *Neurofibromatosis. Conference statement. National Institutes of Health Consensus Development Conference. Arch Neurol 1988;45:575-8.*
2. *Ji J, Shimony J, Gao F, McKinstry RC, Gutmann DH. Optic nerve tortuosity in individuals with neurofibromatosis type 1. Pediatr Radiol 2013;43:1336-43.*
3. *DiMario FJ, Ramsby G, Greenstein R, Langshur S, Dunham B. Neurofibromatosis type 1: magnetic resonance imaging findings. J Child Neurol 1993;8:32-9.*
4. *Listernick R, Charrow J, Greenwald MJ, Esterly NB. Optic gliomas in children with neurofibromatosis type 1. J Pediatr 1989;114:788-92.*
5. *Armstrong GT, Localio AR, Feygin T, et al. Defining optic nerve tortuosity. Am J Neuroradiol 2007;28:666-71.*
6. *Wang MTM, Bhatti MT, Danesh-Meyer HV. Idiopathic intracranial hypertension: pathophysiology, diagnosis and management. J Clin Neurosci Off J Neurosurg Soc Australas 2022;95:172-9.*
7. *Basimah A, Faro SS, Yuan H, et al. Imaging hallmarks of idiopathic intracranial hypertension and insights into pathogenesis. Front Radiol 2025;5:1605777.*
8. *Kwee RM, Kwee TC. Systematic review and meta-analysis of MRI signs for diagnosis of idiopathic intracranial hypertension. Eur J Radiol 2019;116:106-15.*
9. *Pribojszki M, Komáromi K, Csomor A, Vörös E. Tortuous optic nerve. ECR 2017 EPOS. 1 March 2017. Accessed 20 October 2025. Available from: https://epos.myesr.org/poster/esr/ecr2017/C-2914*
10. *Pircher A, Berberat J, Remonda L, Roberts CJ, Neutzner A, Killer HE. Incidence of optic nerve kinking in a cohort of patients with Normal tension glaucoma. Eye (Lond) 2025;39:1270-5.*
11. *Scott RA, Tarver WJ, Brunstetter TJ, Urquieta E. Optic nerve tortuosity on earth and in space. Aerosp Med Hum Perform 2020;91:91-7.*
12. *Curtin BJ. The posterior staphyloma of pathologic myopia. Trans Am Ophthalmol Soc 1977;75:67-86.*
13. *Ehongo A. Understanding posterior staphyloma in pathologic myopia: current overview, new input, and perspectives. Clin Ophthalmol 2023;17:3825-53.*
14. *Liu T, Wang YX, Jonas JB, Hoang QV, Girard MJA, Wang X. Gaze-induced optic nerve head deformations are greater in high myopia and strains increase with axial length. Invest Ophthalmol Vis Sci 2025;66:21.*
15. *Top Karti D, Karti O, Koç AM, Çelebisoy N. Optic nerve tortuosity in neurofibromatosis type 1: a rare case report. Neuroophthalmology 2020;45:313-6.*