NEURO-OPHTHALMOLOGY
2021, VOL. 45, NO. 5, 313–316
https://doi.org/10.1080/01658107.2020.1740282

Taylor & Francis
Taylor & Francis Group

CASE REPORTS

Check for updates

Optic Nerve Tortuosity in Neurofibromatosis Type 1: A Rare Case Report

Dilek Top Karti[a], Omer Karti[b], Ali Murat Koç[c], and Neşe Çelebisoy[d]

[a]Department of Neurology, Bozyaka Training and Research Hospital, İzmir, Turkey; [b]Department of Ophthalmology, İzmir Democracy University, İzmir, Turkey; [c]Department of Radiology, Bozyaka Training and Research Hospital, İzmir, Turkey; [d]Department of Neurology, Ege University, İzmir, Turkey

ABSTRACT

Optic nerve tortuosity is a benign and usually asymptomatic optic nerve abnormality documented on magnetic resonance imaging. This abnormality has rarely been defined in neurofibromatosis type 1 (NF1) cases. In this case report, we present incidental optic nerve tortuosity in a housewife with NF1 without any ocular involvement. Optic nerve tortuosity may occasionally be an incidental finding in patients with NF1. Therefore, clinicians should be aware of this rare coexistence.

ARTICLE HISTORY
Received 15 January 2020
Revised 12 February 2020
Accepted 5 March 2020

KEYWORDS
Genetic abnormality; neurofibromatosis type 1; optic nerve tortuosity

Introduction

Neurofibromatosis type 1 (NF1) is the most common phakomatosis.[1–3] The prevalence is roughly one case per 3000 individuals.[2,3] Approximately half of NF1 cases are inherited as an autosomal dominant trait with complete penetrance and variable expressivity. The remaining cases are associated with de novo germline mutations in the NF1 gene located on chromosome 17.[4,5] This genetic abnormality causes tumour growths on neural tissue that may affect the nervous system, eyes, skin and other parts of the body.[1] Plexiform neurofibromas, Lisch nodules, glaucoma, and optic nerve pathway gliomas are some of its various ocular manifestations.[1] However, optic nerve tortuosity may rarely be the first ophthalmic manifestation of NF1 without the above-mentioned features. We herein present a 24-year-old female with incidentally detected optic nerve tortuosity without any other ocular involvement who had been diagnosed with NF1 ten years earlier.

Case report

A 24-year-old female was referred to our clinic for investigation. On assessment, her only complaint was that of brown flat patches on the trunk of many years' duration. There was a positive family history; her father had suffered from NF1. There was no significant past medical history except for NF1, first diagnosed at the age of 14 years. On systemic examination, her weight and height were within normal limits. There were multiple café-au-lait spots of varying size on the trunk and axillary freckling, but no neurofibromas (Figure 1). The rest of the physical and neurological examination was unremarkable. On neuro-ophthalmological examination, both direct and indirect pupillary light reflexes were normal and no relative afferent pupil defect was observed. Visual acuity and colour vision were normal in both eyes. Examination of the anterior segment and fundus was unremarkable (Figure 2). Intraocular pressure was within normal limits in each eye. Formal visual field testing was normal in both eyes. A comprehensive, magnetic resonance imaging (MRI) examination was performed with a 1.5T machine (Siemens Magnetom Aera, Siemens Healthcare, Erlangen, Germany). T2W axial, T2-STIR coronal and T1W pre/post contrast axial images were acquired all at 3 mm thickness. Phase-contrast 3D venography was also performed afterwards. MRI revealed prominent asymmetrical tortuosity, kinking and thickening of the intraorbital section of the right optic nerve compared with the contralateral side. The optic nerve sheath was preserved bilaterally, but asymmetrically enlarged on the right. The optic nerve signal was normal on axial T2W (Figure 3a) and coronal T2-STIR (Figure 3b) images; no contrast enhancement was observed (Figure 3c). MRI-venography was normal,

CONTACT Omer Karti kartiomer@gmail.com Department of Ophthalmology, İzmir Democracy University, İzmir, Turkey
© 2020 Taylor & Francis Group, LLC

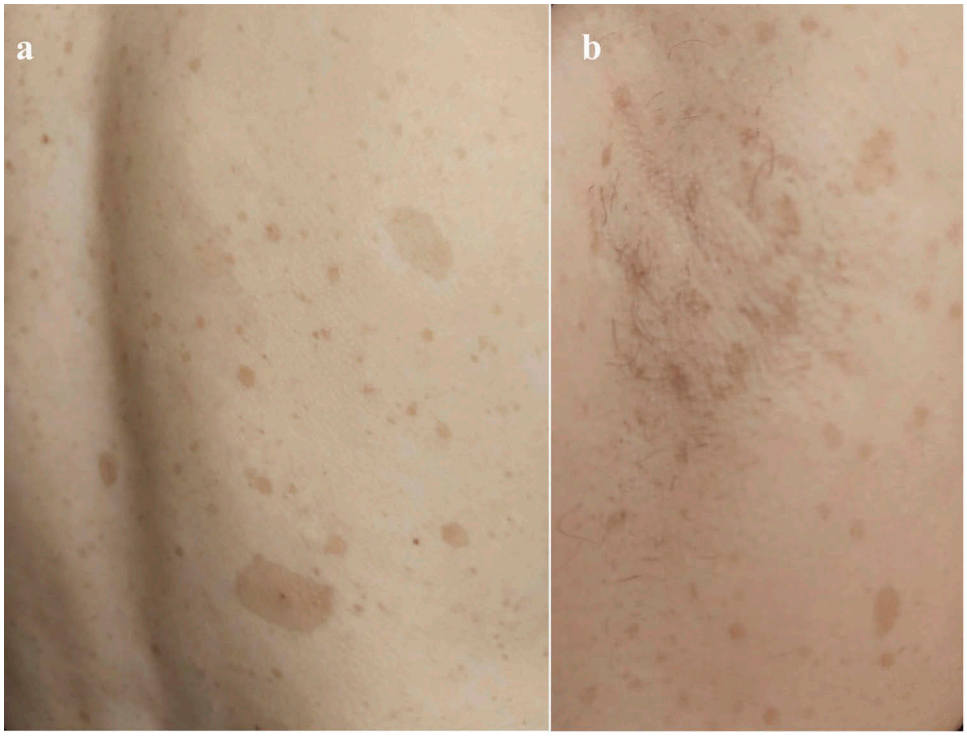


Figure 1. Patient's body photographs showing multiple café-au-lait spots of varying size on the trunk (a) and axillary freckling (b).

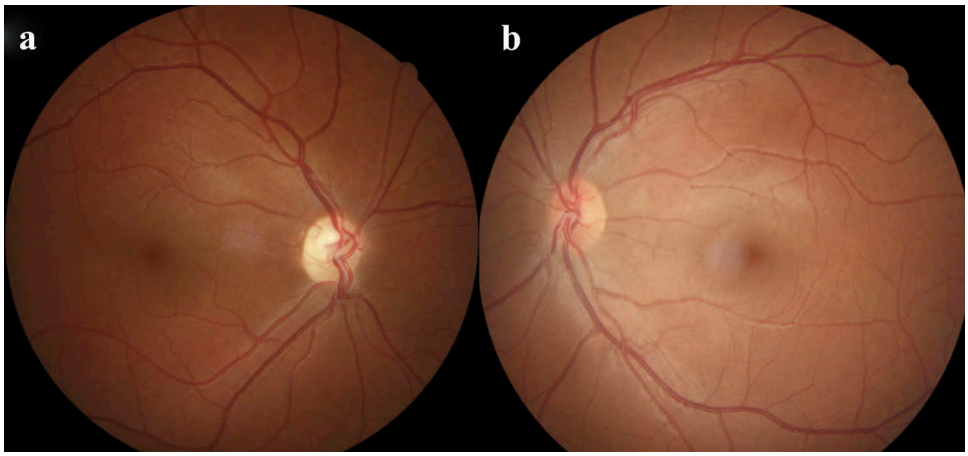


Figure 2. Colour fundus photographs illustrating a normal appearance in the right (a) and left eye (b). (There is no skew deviation of the eyes. The elevated and rotated disk appearance is related to technical reasons).

ruling out a possible cause of elevated intracranial pressure. The absence of contrast enhancement, lack of progression when compared with previous imaging, and lack of the clinical signs of an optic nerve disorder and intracranial hypertension, reliably excluded the diagnoses of optic pathway glioma or pseudotumor cerebri. A diagnosis of non-neoplastic optic nerve tortuosity due to dural ectasia of the optic nerve sheath associated with NF1 was therefore made. Periodic follow-up for the detection of early signs of an optic pathway glioma or other ocular and/or systemic involvement associated with NF1 was recommended.

Discussion

Optic nerve tortuosity is a rare asymptomatic radiological manifestation of a non-malignant optic nerve abnormality that is visible on MRI. Dilatation of the subarachnoid space surrounding the anterior

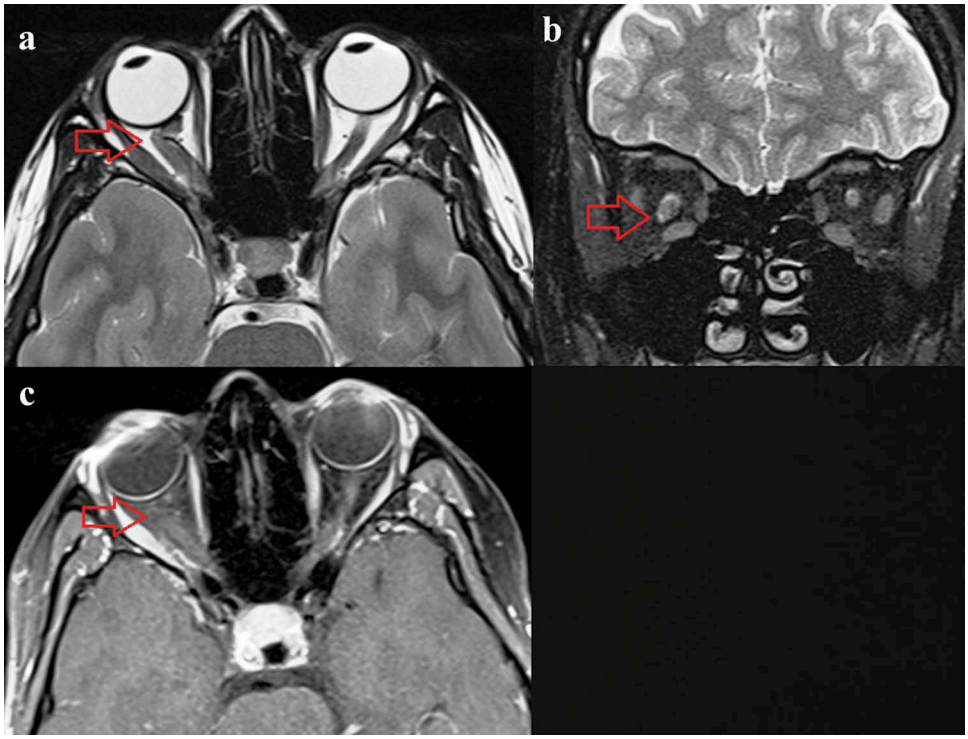


Figure 3. Orbital MRI revealing prominent asymmetrical tortuosity, kinking and thickening of the intraorbital section of the right optic nerve (red arrows) with normal optic nerve signal on axial T2W (a) and coronal T2-STIR (b) images, and lack of contrast enhancement (c). The maximum diameters of the optic nerve and optic nerve sheath were measured in the coronal plane (right: 3.63 mm and 1.97 mm; left 3.56 mm and 1.12 mm respectively).

portion of the optic nerve has been defined by Armstrong et al. as the most reliable feature of optic nerve tortuosity, with high sensitivity and specificity.[6] Besides being detected in healthy individuals as an incidental radiological finding, the abnormality may also accompany some disorders, such as idiopathic intracranial hypertension and NF1.[6,7] The incidence and clinical significance of optic nerve tortuosity in NF1 is unknown. It is also controversial whether optic nerve tortuosity is a precursor of a developing optic nerve glioma. Some studies have reported the relationship between optic glioma and optic nerve tortuosity and suggested that the risk of developing optic glioma may increase in NF1 cases with optic nerve tortuosity at baseline.[6,8] Ji et al.[9] reported that children with NF1 had higher average optic nerve tortuosity than a similar population without NF1. This abnormal morphological appearance of the optic nerve in NF1 cases has been detected irrespective of clinical symptoms or glioma.

In conclusion, optic nerve tortuosity may rarely be seen in NF1 cases. Clinicians, therefore, should be aware of this rare association. In addition, NF1 cases with optic nerve tortuosity should be screened for optic nerve glioma at routine intervals.

Declaration of interest statement

All authors contributed significantly to the creation of this manuscript and each fulfilled criterion as established by the ICMJE. No potential conflict of interest was reported by the authors. Patient consent was obtained to write this case report.

References

1. Abdolrahimzadeh B, Piraino DC, Albanese G, Cruciani F, Rahimi S. Neurofibromatosis: an update of ophthalmic characteristics and applications of optical coherence tomography. *Clin Ophthalmol*. 2016;10:851–860. doi:10.2147/OPTH.S102830.
2. Woodward LM. Ocular manifestations of phakomatoses. *Int Ophthalmol Clin*. 2014;54(3):105–110. doi:10.1097/IIO.0000000000000037.
3. Friedman JM. Epidemiology of neurofibromatosis type 1. *Am J Med Genet*. 1999;89(1):1–6. doi:10.1002/()1096-8628.
4. Cawthon RM, Weiss R, Xu GF, et al. A major segment of the neurofibromatosis type 1 gene: cDNA sequence, genomic structure, and point mutations. *Cell*. 1990;62(3):609. doi:10.1016/0092-8674(90)90253-B.

5. Nandigam K, Mechtler LL, Smirniotopoulos JG. Neuroimaging of neurocutaneous diseases. *Neurol Clin*. 2014;32(1):159–192. doi:10.1016/j.ncl.2013.07.003.
6. Armstrong GT, Localio AR, Feygin T, et al. Defining optic nerve tortuosity. *AJNR Am J Neuroradiol*. 2007;28:666–671.
7. Viglianesi A, Messina M, Chiaramonte R, et al. An interpretation of optic nerve tortuosity. A case report. *Neuroradiol J*. 2009;22:381–385.
8. Levin MH, Armstrong GT, Broad JH, et al. Risk of optic pathway glioma in children with neurofibromatosis type 1 and optic nerve tortuosity or nerve sheath thickening. *Br J Ophthalmol*. 2016;100(4):510–514. doi:10.1136/bjophthalmol-2015-306958.
9. Ji J, Shimony J, Gao F, McKinstry RC, Gutmann DH. Optic nerve tortuosity in children with neurofibromatosis type 1. *Pediatr Radiol*. 2013;43(10):1336–1343. doi:10.1007/s00247-013-2694-1.