Photo essay

Leber's Hereditary Optic Neuropathy: A Case Misinterpreted As Optic Neuritis

Dhaivat Shah, Tina Damani, Shirali Gokharu, Akshar Soni
Choithram Netralaya, Shriram Talawadi, Dhar Road, Indore, Madhya Pradesh, India.

Abstract *A 38 year old male presented with bilateral complete loss of vision over 1 year, left followed by right eye. He was diagnosed to have optic neuritis and was given treatment accordingly, with no improvement in vision. After clinical examination and MRI testing, we diagnosed him to have LHON, explained him the guarded course and nature of the disease and advised gene testing. LHON is a diagnosis of exclusion, and often confused with other optic neuropathies. A sound history, astute clinical examination and appropriate testing can diagnose this rare condition.*

Delhi J Ophthalmol 2022; 32; 72-73; Doi http://dx.doi.org/10.7869/djo.749

Keywords: *Leber's Optic Neuropathy; Hereditary; Fundus Photo; Telangiectatic Vessels*

Case Description

A young 38 year old male presented in our OPD complaining of complete painless loss of vision in both eyes (no perception of light). Patient said that the loss started in left eye first before 1 year, for which he was diagnosed to have optic neuritis and was given intravenous Methylprednisolone (IVMP) pulse dosage. The vision did not improve post IVMP and eventually there was complete loss of vision. Two months back, the patient began to have a similar loss of vision in right eye, and he was diagnosed to have optic neuritis and advised IVMP again. The patient came to us for a second opinion. On fundus examination, the right eye showed mild blurring of disc margin (pseudoedema) with fine peripapillary telangiectatic vessels (yellow arrow) (Figure a). Left eye showed optic disc pallor along with mild tortuosity of retinal arterioles (red arrow) (Figure b). FFA showed no leakage at the optic disc, which ruled out an inflammatory etiology. On further digging into the family history, it was noted that the patient's maternal aunt had a similar problem in her late thirties, and she passed away due to a heart problem when she was 42 years of age. Orbital fat-suppressed contrast enhanced MRI showed mild optic nerve enhancement in both eyes. On basis of all these findings and ruling out other causes, the patient was diagnosed to have Leber's hereditary optic neuropathy (LHON). Gene testing was advised to confirm LHON and patient was referred for genetic counseling. The patient and his family were explained regarding the poor visual prognosis and the course and nature of the disease.

LHON is a rare mitochondrial disease often misinterpreted as or confused with other optic neuropathies. A sound history, astute clinical examination and appropriate testing can aid in diagnosis.[1] Usually these patients tend to present in the second-forth decade. Family history is extremely important to elicit. One eye is commonly involved initially. The optic nerve might show pseudo edema along with fine peripapillary telangiectatic vessels with or without tortuosity of retinal arterioles. FFA will show no evidence of leak at optic disc. In today's era, treatment modalities for LHON include nutritional supplements, activators of mitochondrial biogenesis, brimonidine, idebenone and gene and stem cell therapy.[2]

Declaration Of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have

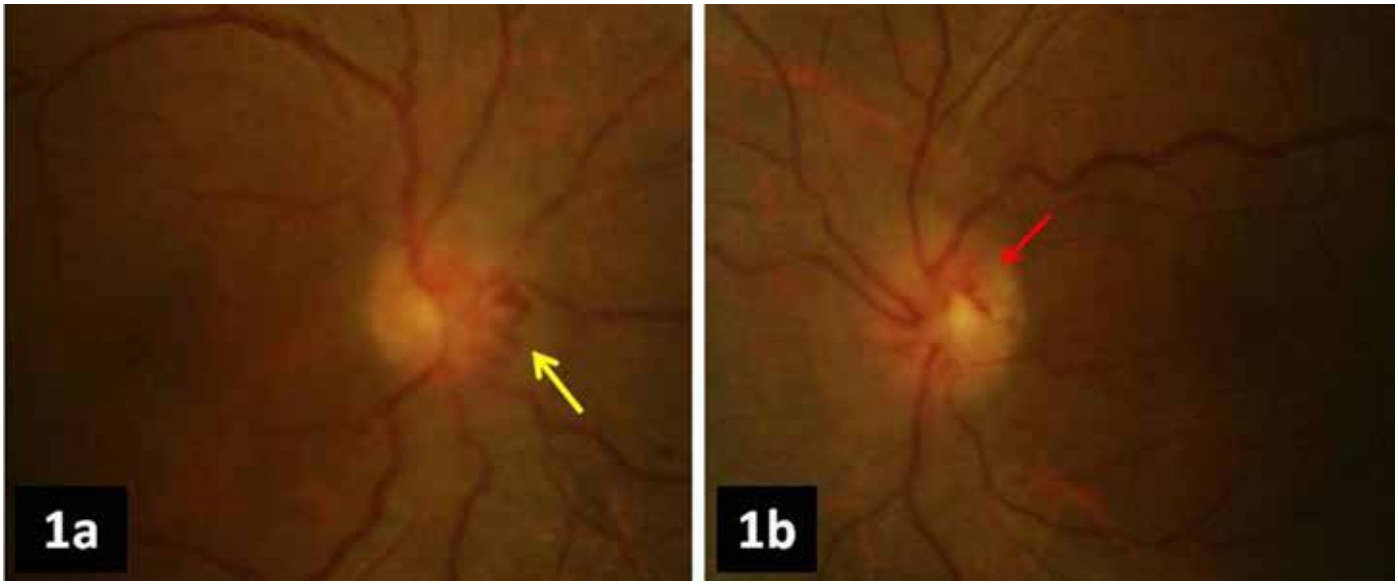

Figure 1: *(1a) Right eye fundus photo showing mild blurring of disc margin (pseudoedema) with fine peripapillary telangiectic vessels (yellow arrow) (1b)Left eye fundus photo showing optic disc pallor along with mild tortuosity of retinal arterioles (red arrow)*

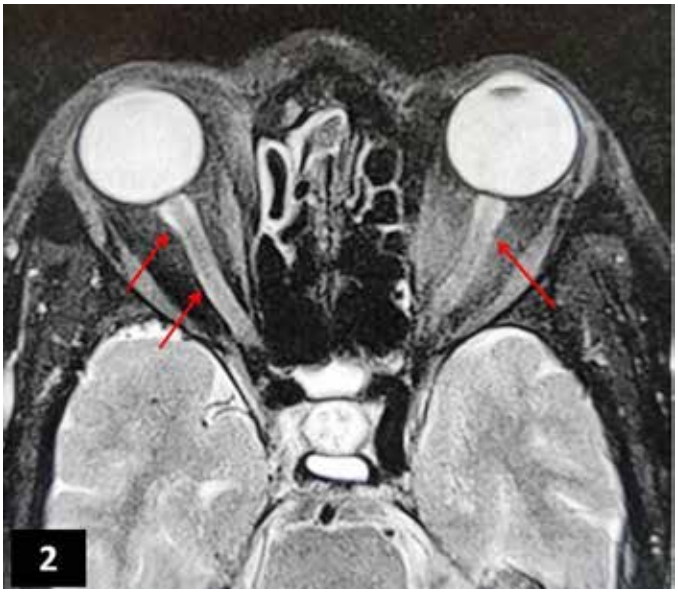

Figure 2: *Orbital fat-suppressed contrast enhanced MRI showing mild optic nerve enhancement in both eyes.*

given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

References

1. Bi R, Logan I, Yao YG. Leber hereditary optic neuropathy: a mitochondrial disease unique in many ways. Handb Exp Pharmacol. 2017;240:309–36
2. Theodorou-Kanakari, A., Karampitianis, S., Karageorgou, V. et al. Current and Emerging Treatment Modalities for Leber's Hereditary Optic Neuropathy: A Review of the Literature. Adv Ther 35, 1510–1518 (2018)

Cite This Article as: Dhaivat Shah, Tina Damani, Shirali Gokharu, Akshar Soni. Leber's hereditary optic neuropathy: A case misinterpreted as optic neuritis . Delhi J Ophthalmol 2022; 32 (3): 72 - 73.

Acknowledgments: Nil

Conflict of interest: None declared

Source of Funding: None

Date of Submission:24 Sep 2020
Date of Acceptance: 26 Jan 2022

Address for correspondence

Dhaivat Shah MS DNB
Deppartment of Opthalmology
Choithram Netralaya, Shriram
Talawadi, Dhar Road, Indore
Madhya Pradesh, India.
E-mail: dhaivatkshah@gmail.com

Quick Response Code