QUESTION 44

How Do I Recognize Leber Hereditary Optic Neuropathy?

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The patient is a 22-year-old man in college who went out drinking over the weekend with his buddies. On Monday, he noted a loss of vision in the right eye to counting fingers. He was found to have a right relative afferent pupillary defect and the nerve looked swollen on the right. A visual field examination was performed and showed a dense central scotoma on the right; the left was normal. Optic neuritis was initially suspected, and he was scheduled for magnetic resonance imaging (MRI) later in the week. Now, however, he has called to say that he has vision loss in his left eye as well. Interestingly, the same thing happened to his brother at age 25 years, a year earlier, and his vision never really recovered. Any ideas?

This young man has suffered severe central visual loss in the right eye. The right relative afferent pupillary defect, the dense central scotoma, and the abnormal-appearing right optic nerve all led you to the appropriate localization of the problem to the right optic nerve. It was most certainly reasonable, in a person of this age with an acute unilateral optic neuropathy, to consider the possibility of inflammation of the optic nerve (ie, optic neuritis). However, if there was no pain at the time of visual loss, optic neuritis would be much less likely (over 90% of patients with optic neuritis will experience pain, usually on eye movement). Similarly, since optic neuritis tends to affect females far more often than males, I am always a bit wary of making a definitive diagnosis of typical idiopathic demyelinating optic neuritis in a male, especially when there is no complaint of pain. The real spoiler in this case is the loss of vision in the other eye within the week. Although optic neuritis can present in both eyes (and indeed the Optic Neuritis Treatment Trial showed that concurrent involvement in the fellow eye was not at all unusual, occurring in more than 40% of patients), involvement of the second eye broadens the differential diagnosis to include infectious and other inflammatory etiologies of optic neuritis (such as syphilis, sarcoidosis, and neuromyelitis optica), as well as other disease categories including rapidly infiltrating neoplasms (such as the leukemias, lymphomas, and malignant optic nerve gliomas), compressive lesions with rapid expansion, such as pituitary apoplexy, or even vascular etiologies. However, the clinical presentation is most suggestive of Leber hereditary optic neuropathy (LHON), and the history of a brother who suffered similar visual loss at age 25 years essentially clinches the diagnosis.

Lee AG, ed.
Curbside Consultation in Neuro-Ophthalmology: 49 Clinical Questions, Second Edition (pp 187-189).