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1 The Diagnosis of Optic Neuropathies

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Abstract
Optic neuropathy refers to disease or damage to the optic nerve, typically characterized by decreased visual acuity, decreased color vision, visual field defect, relative afferent pupillary defect, optic disc edema, and/or optic disc atrophy. Possible etiologies include hereditary, inflammatory, infiltrative, ischemic, demyelinating (optic neuritis), toxic, and compressive optic neuropathies. This chapter discusses the clinical pathway for evaluating and diagnosing optic neuropathy, reviewing the literature in detail.

Keywords: optic neuropathy, hereditary optic neuropathy, radiation optic neuropathy, compressive optic neuropathy, toxic/nutritional optic neuropathy

1.1 Introduction

The diagnosis of an optic neuropathy is usually made on clinical grounds alone. Several excellent references discuss in detail the anatomy of the optic nerve as well as examination techniques.[1,2,3] The clinical features of optic neuropathies include decreased visual acuity, decreased color vision, visual field defect, relative afferent pupillary defect (RAPD), optic disc edema, and/or optic disc atrophy. Other more complex (and time-consuming) testing for optic neuropathy, such as visual evoked potentials (VEPs), flicker fusion, formal color vision testing, and contrast sensitivity, can be performed but in general are not required to establish the clinical diagnosis of optic neuropathy and are not discussed here.

Once the diagnosis of optic neuropathy has been made, it is important to consider a wide differential diagnosis of possible etiologies, including hereditary, inflammatory, infiltrative, ischemic, demyelinating (optic neuritis [ON]), toxic, and compressive optic neuropathies. We refer the reader to the specific chapter on each type of optic neuropathy for further details.

1.2 Can the Appearance of the Optic Nerve Differentiate Etiology?

In general, the appearance of the optic nerve (e.g., normal, swollen, or pale) is not specific and cannot differentiate among various possible etiologies for optic neuropathy. Trobe et al[4] reviewed 163 color fundus photographs of several entities resulting in optic atrophy, including glaucoma, central retinal artery occlusion (CRAO), ischemic optic neuropathy (ION), ON, hereditary optic neuropathy (Leber and non-Leber types), compressive optic neuropathy (CON), and traumatic optic neuropathy (TON). These photographs were reviewed by five ophthalmologists as "unknowns." Glaucoma, CRAO, and ION were correctly identified as the etiology by at least one of the five observers with an accuracy above 80%, but the remaining etiologies were correctly identified in less than 50% of cases. Helpful features in differentiating the entities included the following:

1. The presence of retinal arteriolar attenuation and sheathing in ischemic lesions (e.g., CRAO or ION).
2. Temporal pallor in entities selectively involving central vision and central visual field with sparing of peripheral visual field (e.g., ON and toxic optic neuropathies).
3. Superior or inferior (sector) optic disc pallor in ION.

Although optic disc cupping was often identified in glaucoma, it was also seen in 20% of cases not associated with glaucoma. Optic disc cupping in glaucoma cases, however, was more profound than in nonglaucomatous cases and greater neuroretinal rim pallor occurred in the nonglaucomatous cases. In patients with glaucoma, there is often absence of at least part of the neuroretinal rim, and the color of the remaining rim is normal. With nonglaucomatous optic neuropathy, rarely is any area of the rim completely absent and the remaining rim is often pale. Interestingly, only 11% of these cases with a known history of papillitis or ION had sufficient clues to identify previous disc swelling.[4]

Another study suggested that optic disc appearance may help differentiate anterior ischemic optic neuropathy (AION) from ON, although there are overlapping features. Optic disc stereographs were reviewed by masked observers (87 AION and 68 ON).[5] Altitudinal disc swelling was more than three times more common in AION than ON, although most discs were diffusely swollen. Most patients with AION had hemorrhages, whereas most ON cases did not. Almost all discs with ON had normal color or were hyperemic; only 35% of discs with AION had pallid swelling. Pallid swelling was so rare in ON, however, that of discs with pallor, 93% had AION. Arterial attenuation was also much more typical of AION. AION was the clinical diagnosis in 82% of cases with altitudinal edema, 81% of cases with disc hemorrhage, 93% of cases with pallid edema, and 90% of cases with arterial attenuation. A pale optic nerve with hemorrhage, regardless of type of edema, always represented AION (100%). A hyperemic optic nerve with hemorrhage represented AION in 82% of cases, but if altitudinal edema was also present, the incidence of AION increased to 93%. Conversely, a normal color optic nerve without hemorrhage represented ON in 91% of cases.

In addition, numerous authors have stressed the localizing value to the optic chiasm or optic tract of a special type of optic atrophy caused by specific involvement of the nerve fiber layer of the nasal and temporal retina, respectively. A lesion of the optic tract may result in "band" (or "bow tie") atrophy in the eye contralateral to the involved optic tract. Band atrophy correlates clinically with loss of vision in the temporal visual field and is due to atrophy of the nerve fibers from the nasal half of the retina that then enter the optic disc nasally and temporally. Band atrophy may be unilateral or bilateral with lesions of the optic chiasm.

Neither the pattern (e.g., central scotoma, arcuate, altitudinal) of ipsilateral visual field impairment nor the severity of visual loss is pathognomonic for any specific optic neuropathy, and virtually any visual field defect may occur with any optic neuropathy.[6] In their report on 35 eyes in 20 patients with CON and 70 eyes in 54 patients with ON, Trobe and Glaser[6] found

central scotomas in 33% of cases of CON (vs. 75% in ON) and felt that a central scotoma could not be used as a differentiating feature between the two entities.

The following sections describe the evaluation of optic neuropathy; this approach is summarized in ▶ Fig. 1.1. We begin with an age-based differential diagnosis of an acute optic neuropathy. Two of the most common causes of acute optic neuropathy are AION and ON. Although there is considerable overlap in their clinical presentation, age can be used as an initial differentiating feature in many cases.[7] In younger patients (< 40 years

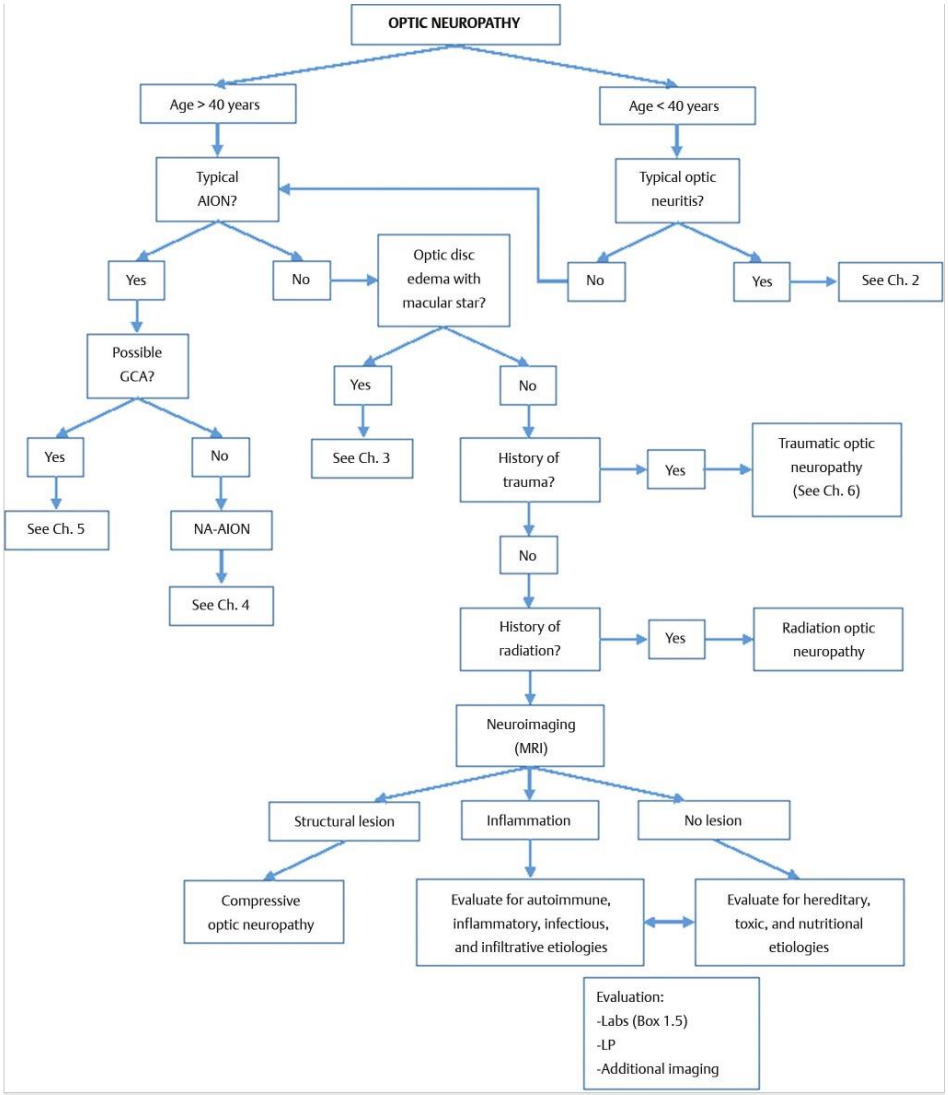

Fig. 1.1 Evaluation of an optic neuropathy. Please refer to the respective chapters on each disorder for additional evaluation and treatment recommendations.