Differential Diagnosis of Optic Nerve Conditions

Comprehensive practitioners must understand various conditions that can adversely affect the optic nerve.

BY MICHAEL McFARLAND, OD

Working in a glaucoma subspecialty clinic, I have come to appreciate the various appearances that an optic nerve can take on, be they acquired, congenital, or physiologic. A thorough stereoscopic optic nerve examination is the first step in making a decision as to whether the optic nerve is healthy or diseased. You must ask yourself, is there optic nerve cupping? Is the nerve pink or pale? Does it appear swollen, or are there distinct margins? Is there a spontaneous venous pulsation? Are there other features such as buried drusen, disc pits, peripapillary atrophy, collateral vessels, hemorrhages, or neovascularization? How large is the disc, and does the cup correlate with this?

The stereoscopic examination coupled with supplementary testing such as visual fields and nerve fiber layer (NFL) analysis will help in making the definitive diagnosis. The presence of a relative afferent pupillary defect more often than not is a sign of a diseased optic nerve. Color vision testing and even something as simple as visual acuity may be helpful in the diagnosis as well. Even with all the testing modalities eye care providers have available in the office, there are certainly times when neuroimaging is needed to rule out the presence of a mass lesion, vessel malformation, or inflammatory process.

IS THERE OPTIC NERVE CUPPING?

If there is abnormal optic nerve cupping along with characteristic visual field and NFL loss, then glaucoma has to be first on the differential diagnosis list. Glaucoma is the only optic nerve condition in which cupping is the primary characteristic appearance.1 It also happens to be the most common optic neuropathy, so it must be ruled out in every case.2 The visual field loss, cupping, and NFL analysis should all correlate nicely with each other. Visual acuity is typically spared until very late in the disease process.1 The remaining part of the rim should be pink in color. If the clinical picture does not make sense, then one must consider other sources of optic nerve disease, such as a previous inflammatory process, ischemic event, or compressive lesion. These conditions may all cause some cupping of the optic nerve, although typically not to the extent that glaucoma does.3-5 Other clues, such as the presence of a pale rim or significantly reduced visual acuity in the absence of retinal disease, will help indicate that the condition is something other than glaucoma.

PINK VERSUS PALE

A pale optic nerve is a sign of atrophy and irreversible optic nerve damage. It typically indicates long-standing optic neuropathy and can be the result of compressive lesions, nutritional/toxic deficiencies, or hereditary disorders. It may also be the end-stage sequela of inflammatory or ischemic events. If the patient is a good historian, background information may be helpful in narrowing the diagnosis. If the visual loss was sudden, it likely indicates an acute inflammatory or ischemic event, such as optic neuritis or ischemic optic neuropathy as the underlying culprit. A more gradual onset of vision loss over a period of months to years suggests a compressive lesion, hereditary condition, or nutritional/toxic deficiency.3

THE SWOLLEN OPTIC NERVE

Key features of the swollen optic nerve are blurred disc margins, hyperemia of the optic nerve head, and an anterior elevation of the optic nerve head. Some causes of a swollen optic nerve include the anterior ischemic optic
neuropathies, central retinal vein occlusion, compressive lesions, increased intracranial pressure, as well as inflammatory and infiltrative diseases. The term papilledema is used when the swelling is secondary to increased intracranial pressure. If the disc is swollen without an associated increase in intracranial pressure, then the term disc edema should be used.6

If an elderly patient presents with a swollen optic nerve along with complaints of headache, neck pain, jaw claudication, weight loss, and/or chronic fatigue, temporal arteritis must be ruled out immediately, as this is a true ophthalmic emergency.3 Buried optic nerve drusen can sometimes mimic disc edema.2 The presence of irregular and scalloped disc margins can signal the presence of disc drusen. Ocular coherence tomography (OCT), fundus autofluorescence, ultrasound, and even a computed tomography scan may be helpful if there is any question as to whether the practitioner is looking at a swollen nerve or disc drusen.

VISUAL FIELD AND OCT TESTING

Visual field testing is an essential component in the diagnosis and management of optic neuropathy. Various patterns of visual field loss include arcuate scotomas, altitudinal defects, central scotomas, and generalized depression. No one pattern is specific for a particular optic neuropathy, but there are clear trends that will help narrow the diagnosis. Altitudinal defects are often caused by ischemic optic neuropathies, while cecocentral scotomas are more commonly associated with hereditary conditions, nutritional deficiencies, or due to toxicity. Enlarged blind spots may occur in cases of papilledema secondary to idiopathic intracranial hypertension. A hemianopic defect indicates a lesion at or posterior to the optic chiasm.1,6 It is imperative for the practitioner to be familiar with the most common types of visual field loss and the conditions that cause them. Visual field analysis is an excellent tool not only in the diagnosis of the condition, but also in determining the extent of nerve damage and following the condition over time.

The OCT’s ability to rapidly and accurately measure ocular parameters such as NFL thickness and optic nerve morphology clearly make it a valuable tool in the diagnosis and management of optic nerve pathology. It is very useful in detecting which part of the optic nerve is damaged and allows the eye care provider to correlate this with the rest of the clinical findings. It may be effectively used in some cases to differentiate borderline cases of papilledema and is a good adjunct to stereoscopic nerve evaluation and visual field testing. The ability of OCT to image axons has garnered great attention in its use in following and aiding in the diagnosis of patients with multiple sclerosis with and without a history of optic neuritis.8 Newer OCT software has the ability to quantify the thickness of the ganglion cell layer in the macula, allowing clinicians to further appreciate the extent that optic nerve disease may have on visual acuity and contrast sensitivity.

CASE STUDY

A 43-year-old man was referred to the glaucoma service for a second opinion on his management. He had been placed on glaucoma medications by the referring doctor, but was unable to tolerate the drops. His intraocular pressure was 12 mm Hg in both eyes off all glaucoma medications. The BCVA was 20/25 in his right eye and 20/100 in the left. Color vision was reduced in the left eye only. Visual field testing revealed a superior arcuate defect in the right eye along with some inferior depressed points. The left eye showed a dense superior arcuate defect along with an inferior temporal defect that appeared to respect the vertical midline. Foveal sensitivities were 35 dB in the right eye and 0 dB in the left eye. The OCT scan demonstrated questionable early retinal NFL loss in both eyes. The patient’s cup-to-disc ratio was graded as a 0.4 in both eyes with no edema or pallor noted.
The combination of reduced visual acuity, color vision loss, and significant visual field defects in the absence of significant cupping warranted further evaluation with neuroimaging (Figure). A magnetic resonance image of the brain and orbits revealed a 16-mm pituitary mass impinging on the optic chiasm consistent with a macroadenoma. The patient was referred to a neurosurgeon for further evaluation and treatment.

CONCLUSION

When diagnosing optic neuropathies, things need to make sense. Glaucoma should be high on the considered differential diagnosis, but as clearly indicated in the case example, this is not the only issue one should consider. There were multiple factors that seemed to indicate that the patient had something other than glaucoma, but still the diagnosis was missed. As comprehensive practitioners, it is mandatory that we understand and appreciate the various conditions that can adversely affect the optic nerve and other parts of the visual system. Slowing down and making sure that your clinical examination correlates with the supplementary testing will make for a more effective practitioner.

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