

A Case of Legal Blindness Secondary to Optic Nerve Head Drusen

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Abstract

Optic nerve head drusen are relatively infrequent abnormalities of the optic disc that are usually discovered incidentally during a routine eye examination. While drusen are typically considered to be benign, visual field loss due to drusen is not uncommon. Visual field defects are more prevalent in superficial drusen than buried drusen, and can present as nerve fiber bundle defects, generalized concentric constriction, or enlargement of the blind spot. The extent of damage is variable, but the degree of field loss rarely leads to substantial functional impairment. This case report presents a patient with significant bilateral concentric field loss that was presumed to be secondary to optic nerve drusen that caused severe visual impairment and disability.

KEY WORDS:

optic nerve head drusen, legal blindness, visual field, low vision, optical coherence tomography

INTRODUCTION

Optic nerve head drusen (ONHD) are extracellular calcific deposits that occur in small, crowded optic nerve heads. While the pathogenesis is poorly understood, they are believed to result from a defect in axoplasmic transport and axonal degeneration of optic nerve fibers.¹⁻³ Abnormal patterns in the disc vessels combined with a narrow scleral canal may make the nerve fiber more prone to develop intracellular mitochondrial calcifications; axons then rupture and extrude debris into the extracellular space anterior to the lamina cribrosa and posterior to Bruch's membrane. These deposits act as a nidus for more calcium accumulation, causing the drusen to increase in size over time.¹⁻³

ONHD are typically found incidentally during a routine eye exam, and patients are often asymptomatic.^{2,3} Although ONHD can be associated with other ocular conditions such as angioid streaks, pseudoxanthoma elasticum, and retinitis pigmentosa, they are usually isolated non-syndromic abnormalities.² The prevalence ranges from 0.34% to 2.4%; they are more common in females than males, and are bilateral in 65% to 90% of cases.^{1,2} ONHD develop in the first decade of life, but are buried in the nerve head and may not be visible on examination in their early stages. These nerve head concretions tend to slowly progress with age and become more superficial, manifesting as rounded, refractile deposits within the nerve and causing an irregular surface appearance with indistinct disc margins.^{4,5}

Visual field loss is common with ONHD, with a wide reported range of from 24% to 87%.² These field defects vary widely in the degree of their severity, and in rare circumstances can lead to significant visual impairment.⁶⁻⁹ Legal blindness is defined in both the United States and Canada as a best corrected visual acuity of 20/200 or less in the better eye, or a visual field constriction of 20 degrees or less.^{10,11} Although it is not a true indication of functional status, this classification is used to determine eligibility for programs such as vocational training, rehabilitation, and tax exemp-

tion status. The following case report presents an unusual instance of severe visual field loss causing legal blindness in an older patient with ONHD.

CASE REPORT

A 68-year-old male Vietnam war veteran with a known history of dry macular degeneration and ONHD in both eyes was examined after transferring from another facility where he had been followed by a retina specialist. He had been a helicopter pilot while on active military duty and reported having no visual difficulties during that time. For several years he had noticed more difficulty with mobility; he felt like his peripheral vision was getting worse, he was running into obstacles while walking, and had given up driving. His medical history was significant for benign prostate hypertrophy, chronic obstructive pulmonary disease, hyperlipidemia, hearing loss, chronic post-traumatic stress disorder, myasthenia gravis, chronic back pain with peripheral neuropathy, and depression.

The corrected distance visual acuities were 20/25 OD and 20/30 OS. Slit lamp examination found moderate nuclear cataracts in both eyes, and the pupils were round and reactive with no relative afferent defect. The intraocular pressures were 16mmHg OU by Goldmann applanation tonometry. Dilated fundus examination showed a superficial, irregular elevated contour to the rim tissue of the optic nerve, with irregular disc margins, obscuration of the physiologic cup, and peripapillary atrophy of both nerves (Figure 1). Mild disruption of the retinal pigment epithelium and soft drusen were noted in the macula of both eyes.

B-scan ultrasound demonstrated a hyperechographic density on both optic nerve heads with high and low gain settings, consistent with the history of ONHD (Figure 2). Spectral domain optical coherence tomography (SD-OCT) also showed elevated, lobular, irregular surfaces on both nerve heads (Figure 3), with superior and inferior thinning of the retinal nerve fiber layer (Figure 4). Visual field testing revealed dense bilateral concentric constriction with a small central island of vision remaining in each eye (Figures 5 and 6). Previous magnetic resonance imaging (MRI) scans of the head and orbits with and without contrast did not find any corresponding pathology, and a review of general medical notes and laboratory testing did not reveal other contributing factors. The patient was classified as legally blind based on the field loss, with nuclear cataracts and early age-related macular degeneration contributing to the decreased visual acuity.

After a discussion of treatment options, the patient declined the use of topical intraocular pressure-lowering medications to reduce the risk of further progression of the visual field loss. Low vision rehabilitative services were provided at both the local facility and a regional Blind Rehabilitation Center, where he was issued multiple low vision devices and received orientation and mobility training and computer access training. He continued periodic exams with repeat vi-

sual field and SD-OCT testing without progression of the visual field loss or change in the retinal nerve fiber layer over the next 10 years. Uncomplicated cataract surgery was eventually performed in both eyes, but significant deterioration of the macular degeneration over the next decade caused a further reduction of visual acuity. The macular degeneration remained dry OS, but a choroidal neovascular membrane OD was treated by a retina specialist with several injections of bevacizumab (Avastin) and aflibercept (Eylea). At his last examination, the corrected distance visual acuities were 20/60 OD and 20/800 OS.

DISCUSSION

It is critical to distinguish ONHD from true papilledema due to increased intracranial pressure, which is a medical emergency with potential life-threatening consequences. Both conditions cause a bilateral elevated appearance of the optic nerve with indistinct margins, and both sometimes report bilateral transient visual obscurations. The optic nerves in papilledema may be hyperemic with flame-shaped hemorrhages, although hemorrhage can occasionally be noted in ONHD. In contrast to ONHD, the adjacent nerve fiber layer will be swollen with papilledema, often obscuring the underlying blood vessels. Other clinical signs of papilledema can include cotton wool spots, hyperemia, venous congestion, Paton's lines, and exudates. Papilledema may also be accompanied by systemic neurological symptoms such as headaches, nausea, tinnitus, or vomiting.^{1,3,12}

The diagnosis of visible superficial ONHD is usually straightforward from their characteristic clinical appearance, but additional ancillary testing can help confirm the clinical finding and may be needed to discern deeper-buried drusen as well as help differentiate from papilledema. B-scan ultrasound has been the preferred diagnostic test due to its accuracy and non-invasive nature, and, with good technique, can be effective for imaging both superficial and buried ONHD; the drusen will appear on the scan as discrete areas of high reflectivity at the nerve head which persist at low gain.^{12,13} ONHD can be visualized with computed tomography (CT) due to their calcific composition, but this method is not as reliable as ultrasound due to limitations in resolution with standard-size image slice thickness and variability in axial positioning of the image slices through the optic nerve head.^{12,14} Superficial ONHD will show nodular staining without leakage in late-phase photos on fluorescein angiography; in contrast, true optic disc edema will have diffuse hyperfluorescent leakage in the early phase. Superficial ONHD may also be visualized with fundus autofluorescence as discrete round or oval hyperfluorescent areas with irregular borders. Neither of these techniques are sensitive for detecting buried ONHD.^{12,15} SD-OCT may be valuable in the diagnosis, manifesting as an elevated, irregular surface of the nerve head, with rounded, hyporeflective cavities and hyper-reflective borders, though in some cases it is difficult to distinguish ONHD from large superficial blood vessels.¹⁶⁻¹⁹ Increased thinning of the retinal nerve fiber layer (RNFL) and macular ganglion cell layer are associated with the progression of ONHD as they become more superficial, and SD-OCT can be used to monitor ongoing changes due to ONHD over time.^{12,18,19}

While ONHD are mostly benign and visual acuity tends to be well preserved, ophthalmic side effects can occur. Visual field defects are the most common ocular complication, but retinal hemorrhages and vascular occlusions are reported in 2% - 10% of patients and can cause acute vision loss.^{12,20} Nonarteritic anterior ischemic optic neuropathy, central retinal artery and vein occlusions, retinal nerve fiber hemorrhages, vitreous hemorrhage, and choroidal neovascular membranes have all been associated with ONHD.²⁰⁻²⁴ The pathophysiology is not known, but it is presumed that as the calcific bodies enlarge in the crowded optic disc and small scleral canal, the mechanical stress impairs axoplasmic flow or causes a compressive ischemia which leads to nerve fiber layer damage, vascular compromise, and progressive field loss.²⁵ Though these ophthalmic complications may be due to underlying ONHD, patients with these conditions should still undergo additional testing per established protocols to rule out other standard comorbid risk factors.

ONHD were first described as a cause of visual field defects in 1921, and the association has been well documented since then.²⁶ Visual field defects are significantly more prevalent with superficial ONHD (71% - 75%) than with buried ONHD (21% - 48%), and tend to manifest with older age as the drusen progress and become more superficial over time.²⁶⁻²⁸ The most common types of visual field defects found in eyes with ONHD are nerve fiber bundle defects, generalized concentric constriction, and enlargement of the blind spot, usually with preservation of central visual acuity and central visual field. The rate of progression of the visual field loss is slow, estimated at 1.6% per year, and patients are often unaware that the visual field is getting worse.²⁹ The prevalence of severe visual field loss causing legal blindness is unknown, but it is rare and only occasionally reported in the literature.^{6,9}

The types of visual field loss found in ONHD are also characteristic for primary open angle glaucoma (POAG). In addition, the pattern of RNFL loss found on OCT may be similar to glaucomatous thinning depending on the location of the drusen in the nerve head, further confounding the diagnosis. Small, crowded optic nerves with ONHD may mask glaucomatous structural damage, presenting a diagnostic challenge with comorbid ONHD and POAG as to the etiology of the structural and functional changes. Patients with ONHD and elevated intraocular pressure (IOP) should be monitored more closely as they may be more at risk for the progression of visual field loss, with treatment offered as appropriate if glaucoma is suspected.³⁰

Management options for progressive visual field loss from ONHD are limited, and no definitive treatment is currently available. Lowering IOP with antiglaucoma medication might have a theoretical benefit of alleviating the mechanical compression of ganglion cell axons and improving optic nerve head perfusion, perhaps decreasing the risk of progressive field loss.³¹ Since there have been few studies on the relationship between IOP and visual field defects in ONHD, there is incomplete evidence to support this strategy. One study found that ONHD eyes with concomitant ocular hypertension had a higher risk of visual field loss, and the authors recommended that eyes with ONHD and ocular hypertension be treated to lower IOP.³⁰ Another retrospective chart review demonstrated that higher IOP in normotensive eyes with ONHD was not associated with greater visual field loss or a thinner retinal nerve fiber layer, suggesting that lowering IOP may not prevent visual field loss in normotensive eyes; however, a different, small, short-term prospective investigation treated normotensive ONHD eyes with topical brinzolamide and seemed to show that decreasing IOP improved retinal ganglion cell function and delayed the progression of optic neuropathy.^{32,33} More research is needed to better understand the possible long-term benefits of lowering IOP in ONHD patients. Surgical removal of ONHD has been performed but can result in poor visual consequences, and variability in the drusen consistency and hardness make the results unpredictable.³⁴ Mechanical decompression with radial optic neurotomy has shown some success, but with only a few case series reported it has not been widely adopted.^{35,36}

CONCLUSION

Although ONHD are generally considered a benign condition with a favorable prognosis, some degree of associated visual field loss is common. Most patients remain asymptomatic and the severe visual field loss experienced by the present patient is rare. The diagnosis should be confirmed through clinical exam supplemented by auxiliary testing to rule out other comorbidities. Neuroimaging is recommended in cases of substantial visual field loss to rule out compressive optic neuropathy. Even though progression is generally slow, patients must be followed periodically with dilated exams, serial visual fields and OCT to document any structural or functional changes. While no therapeutic intervention has yet been proven to be effective and most presentations require only observation, medical or surgical management can be discussed in progressive cases, and low vision rehabilitation should be offered in cases of functional impairment. ●

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Figure 1: Fundus photos of optic nerve head drusen OU in a patient with severe visual field loss.

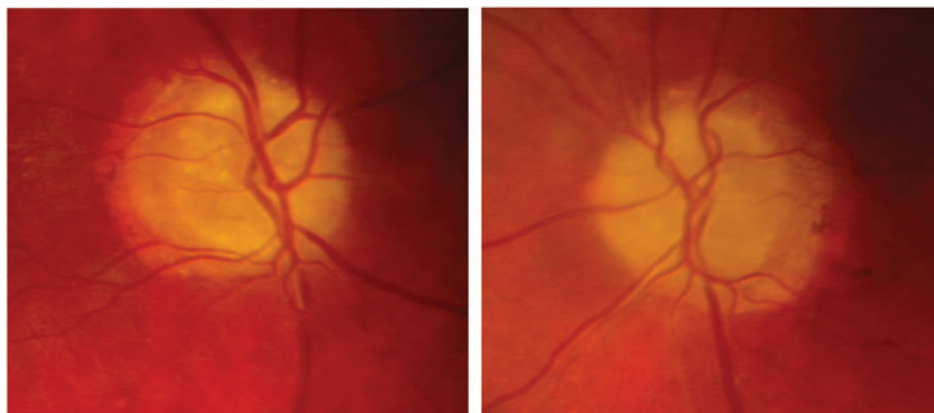


Figure 2: B-scan ultrasound at high gain (Figure 2a) and low gain (Figure 2b) showing hyperechographic densities on the optic nerve heads OU, consistent with optic nerve head drusen.

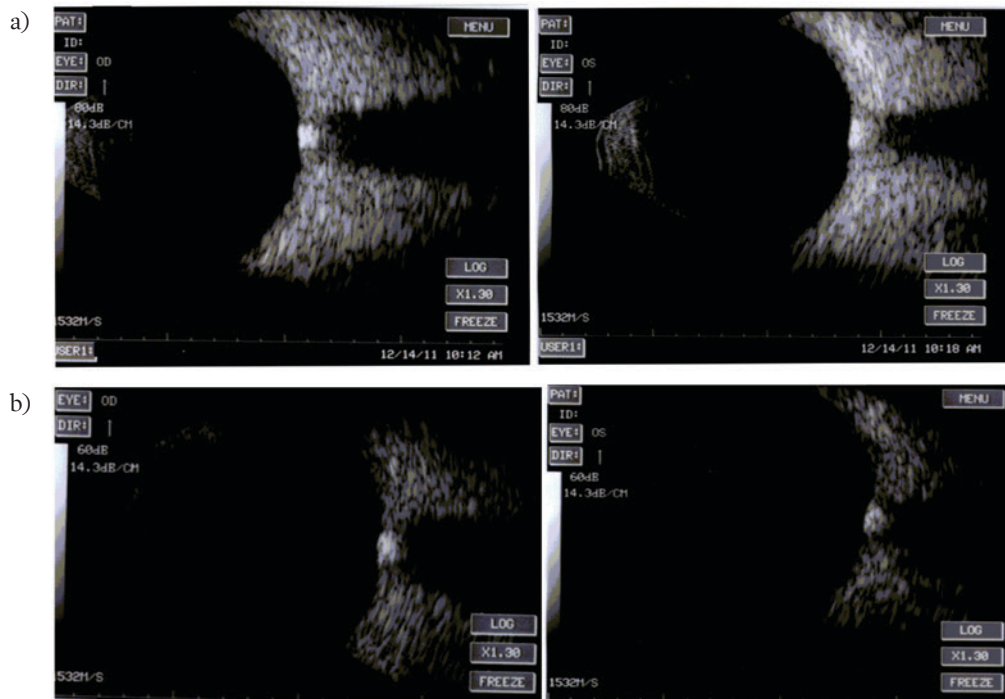


Figure 3: Optical coherence tomography of the optic nerve head drusen shows irregular nerve head surfaces and underlying hyporeflective cavities of both eyes due to optic nerve drusen.

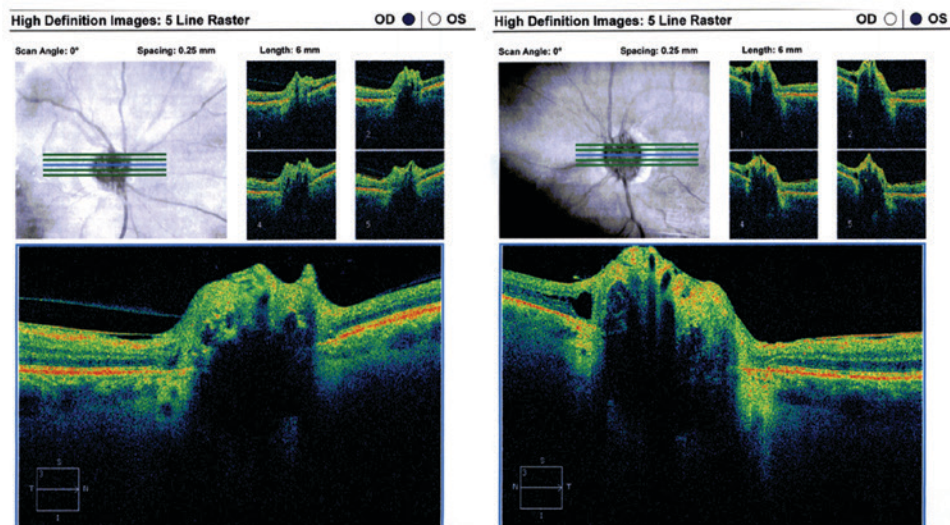


Figure 4: RNFL thinning due to optic nerve drusen was found on optical coherence tomography.

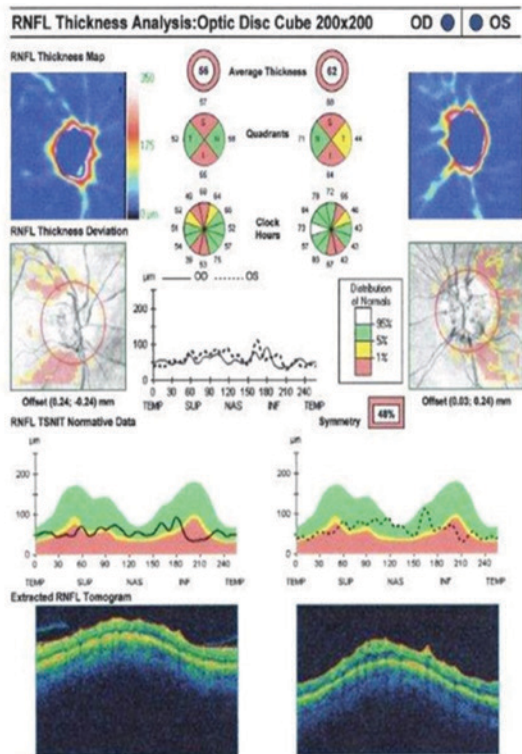


Figure 5: Humphrey visual fields showed severe bilateral constriction due to optic nerve drusen.

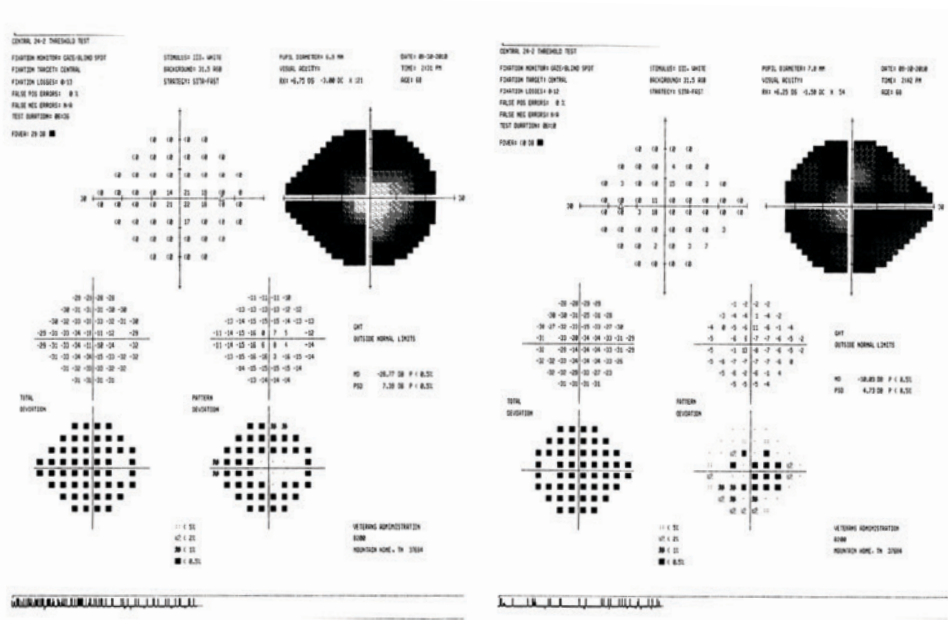
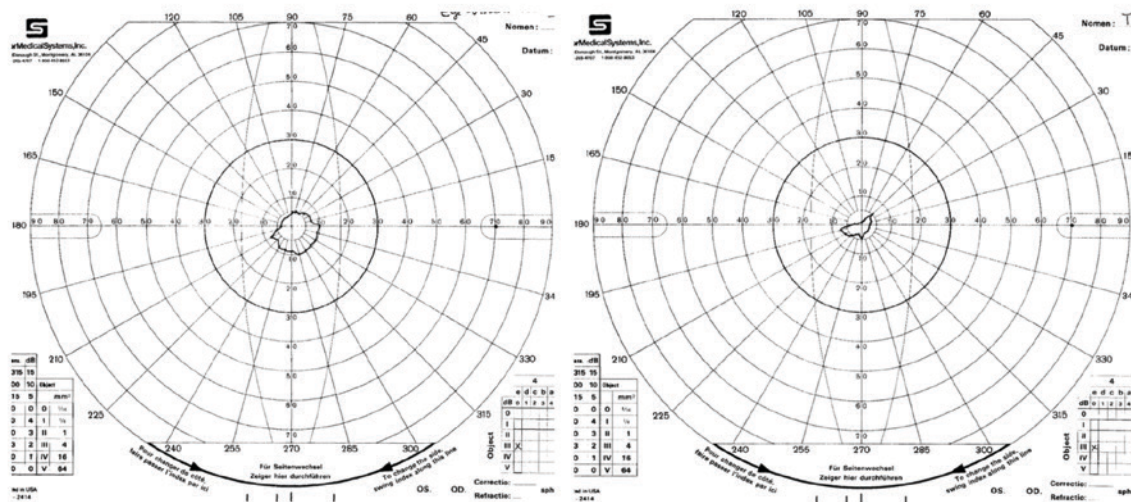


Figure 6: Goldmann visual fields confirmed severe constriction OU due to optic nerve head drusen.



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