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Optic nerve head drusen in black patients

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Abstract

Background—Several studies have suggested racial differences in the prevalence of optic nerve head drusen (ONHD). We aimed to determine the percentage of patients with ONHD who are black, and to describe the clinical, ophthalmoscopic, and perimetry findings in these patients.

Methods—We conducted a retrospective chart review of all patients with ONHD seen at our institution between 1989 and 2010. Only black patients with ONHD confirmed on either funduscopy or B-scan ultrasonography were included. Demographic and clinical findings in these patients were recorded and analyzed.

Results—Of 196 patients with confirmed ONHD, 10 (5.1%) were black (7 women; ages 8–61 years). Six of the 10 patients had bilateral ONHD. The ONHD were buried in 11 of 16 eyes with ONHD, and exposed in 5 of 16 eyes. Fifteen of 16 eyes with ONHD had small cupless optic nerve heads. Visual fields were normal in 4 of 16 eyes with ONHD. In the other eyes, visual field defects included an enlarged blind spot (5 eyes), constricted field (5 eyes), nasal defect (2 eyes), central defect (1 eye), and generalized depression (1 eye). Visual field defects were present in 4 of 5 eyes (80%) with exposed ONHD and 8 of 11 eyes (72.7%) with buried ONHD. None of the patients were related and none of their examined family members had exposed ONHD on funduscopic examination.

Conclusion—ONHD are rare in blacks, possibly due to the presence of a larger cup-to-disc ratio or a lack of predisposing genetic factors. Visual field defects are common in black patients with both exposed and buried ONHD.

Search Terms

Optic Nerve; Drusen; Ethnicity

Introduction

Optic nerve head drusen (ONHD) are laminated acellular concretions that form within the substance of the optic nerve [1]. They often occur in small structurally-congested optic nerve heads and are inherited in an autosomal dominant fashion with incomplete penetrance [2]. Several studies have suggested racial differences in their prevalence [3–6], such that ONHD are said to occur almost exclusively in whites [4]. The prevalence of ONHD in

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blacks is unknown. An autopsy series of American patients found that the prevalence of buried and exposed ONHD was about 2%, but the races of these patients were not specified [7]. A series of American patients from Miami, where a significant proportion of the population is black, reported that only 2 of 98 patients with exposed ONHD on fundus examination were black [4]. A more recent American series, including patients from Galveston (Texas) and Miami, found that 5 of 85 patients with exposed ONHD were black [5]. Since equal proportions of black and white patients are seen at our institution, we performed a retrospective chart review to determine the percentage of patients with exposed or buried ONHD who were black, and to describe the clinical, ophthalmoscopic, and perimetry findings in these patients.

Methods

We retrospectively reviewed the clinic charts of all patients with exposed or buried ONHD seen between 1989 and 2010 at our institution. All patients had received a standardized neuro-ophthalmic assessment, including formal (Humphrey 24–2 or Goldmann) perimetry and fundus photography, and their relatives had been examined whenever possible. The nature of any visual field defect was determined by the clinician at the time of the patient's evaluation and by the author compiling the database (MJT), who was not blinded to the clinician's visual field interpretation. In cases where there was a discrepancy in visual field interpretation, the nature of the visual field defect was determined by consensus among the study authors. Race was determined by the clinician, based on patient appearance. When the clinician was not certain of the patient's race, the patient was asked to report their race. We only included black patients who had ONHD confirmed on either fundus examination or B-scan ultrasonography. We excluded non-black patients and all patients in whom an alternative cause for optic nerve head elevation was identified. The study protocol was approved by the Emory University Institutional Review Board.

Results

Of 196 patients with ONHD on fundus examination or B-scan ultrasonography, 10 (5.1%) were black (7 women; ages 8–61 yrs, mean 25 yrs, median 15 yrs); the demographic and clinical characteristics of these patients are summarized in Table 1. No patient with ONHD was excluded from the study due to the presence of an additional cause for optic nerve head elevation. Seven of the 10 patients had no visual symptoms and had been referred for evaluation of abnormal optic nerve heads. Two of the 3 patients with visual symptoms (patients 3 and 10; see Table 1) complained of progressive visual field loss in both eyes, and the third patient (patient 6; see Table 1) complained of progressive dyschromatopsia in the right eye only. Of the 10 patients, only one (patient 8; see Table 1) had another ophthalmic disease; in this case, the patient had keratoconus in an eye without ONHD. None of the patients reported a sudden loss of vision to suggest anterior ischemic optic neuropathy or central retinal artery occlusion, and none of the patients had symptoms or signs of raised intracranial pressure. None of the patients were related and none of their examined family members had exposed ONHD on fundus examination. Six of the 10 patients had bilateral ONHD. Visual acuity was normal in most eyes with ONHD (range −0.1 to 0.4, mean 0.08, median 0.0; logMAR notation). Intraocular pressures were within normal limits in all eyes with ONHD. Fifteen of 16 eyes with ONHD had small cupless optic nerve heads, 13 eyes had elevated optic nerve heads, and 5 eyes had exposed ONHD. Perimetry was normal in 4 of 16 eyes with ONHD, whereas there was an enlarged blind spot in 5 eyes, a constricted field in 5 eyes, a nasal defect in 2 eyes, a central defect in 1 eye, and generalized depression in 1 eye. Visual field defects were present in 4 of 5 eyes (80%) with exposed ONHD and 8 of 11 eyes (72.7%) with buried ONHD. No alternative cause for the visual field defects (e.g., glaucoma, papilledema, or compressive lesion) was identified in any patient.

Discussion

Several prior studies have suggested racial differences in the prevalence of ONHD, but these studies have only included patients with exposed ONHD on fundus examination or photographs [3–6]. We included patients with exposed or buried ONHD, confirmed on either fundus examination or B-scan ultrasonography, and found that only a small percentage were black, implying that there is a lower prevalence of ONHD in blacks compared to whites. Since approximately 50% of the patients examined at our institution are black, the disproportion is unlikely to be due to referral bias.

There are several possible explanations for the low prevalence of ONHD in black patients. Firstly, racial differences in optic disc morphology may be relevant. It has been proposed that a small scleral canal size may play a role in the pathogenesis of ONHD, since ONHD usually occur in eyes with small cupless optic nerve heads [8]. Indeed, almost all eyes with ONHD in our series had small cupless optic nerve heads. Several studies have demonstrated that, on average, blacks have larger optic disc areas, with larger cups and cup-to-disc ratios, compared with whites [9,10]. Thus, ONHD may be less common in blacks than in whites, due to a larger average scleral canal size. A recent study using optical coherence tomography (OCT) found that the average scleral canal size in eyes with ONHD was not significantly smaller than in control eyes or fellow eyes without ONHD, calling into question whether scleral canal size has any role in the pathogenesis of ONHD [11]. Although the optic nerve heads of the patients in our series appeared small and structurally-congested, OCT measurements of scleral canal size were not available for any of the patients and, thus, we are unable to be certain that scleral canal size was smaller than average in these patients.

A second possibility is that the low prevalence of ONHD in black patients might be due to a lack of predisposing genetic factors. Several previous studies have demonstrated that ONHD are inherited in an autosomal dominant fashion with incomplete penetrance [2,3]. Although a candidate gene has not yet been identified, genes have been identified for other autosomal dominant and recessive disorders in which ONHD can be a feature [12–14]. None of the patients in our series had ONHD as a feature of another disorder. Furthermore, none were related and none of their examined family members had exposed ONHD on fundus examination, suggesting that ONHD may be sporadic rather than an inherited trait in these patients. However, we examined only those relatives who accompanied the patient and we did not systematically arrange for other relatives to be examined. Given the small number of patients and incomplete pedigree information, it therefore remains possible that ONHD might be an inherited trait in these patients.

The clinical and ophthalmoscopic findings in our patients were similar to those reported in other large series of ONHD [2–4]. The percentage of our patients with exposed ONHD who had visual field defects (80%) was slightly greater than that reported in prior studies (~70–75%) [15–18]. However, the percentage of our patients with buried ONHD who had visual field defects (72.7%) was substantially greater than that reported in prior studies (~20–45%) [15–18]. While this could be a consequence of referral bias, it might also indicate that visual field defects are more likely to develop in black than in white patients with ONHD, possibly due to racial differences in the sensitivity of retinal ganglion cells to damage at the optic nerve head.

In summary, we have demonstrated that only a small percentage of patients with ONHD are black, suggesting that there is a low prevalence of ONHD in blacks compared with whites. Although the lower prevalence could be due to a larger average cup-to-disc ratio or a lack of predisposing genetic factors in blacks, a large population-based study, with fundus

examination or photography and B-scan ultrasonography, would be required to determine the exact prevalence of ONHD in blacks.

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Table 1

Demographic and clinical characteristics of black patients with optic nerve head drusen (ONHD)

Patient	Age (yrs)	Sex	VA		RAPD	Optic Nerve Head Appearance	Perimetry [MD]	B-Scan
1	8	F	OD	0.0	–	Small, elevated, exposed ONHD	EBS, nasal [−15.63]	Calcified ONHD
			OS	0.0	–	Small, elevated	Central [−4.18]	Calcified ONHD
2	12	M	OD	0.0	–	Normal	Full [GVF]	Not done
			OS	0.0	–	Elevated, exposed ONHD	Full [GVF]	Not done
3	14	F	OD	0.0	–	Small, elevated	Inf>sup arcuate [−16.03]	Calcified ONHD
			OS	0.0	Yes**	Small, elevated	Inf>sup arcuate [−16.78]	Calcified ONHD
4	14	M	OD	0.2	–	Small, elevated	EBS, nasal [−4.48]	Calcified ONHD
			OS	0.4	–	Small, elevated	Full [−2.49]	Calcified ONHD
5	15	F	OD	0.0	–	Small, elevated	Full [−1.83]	Calcified ONHD
			OS	0.0	–	Small	Full [−2.94]	Normal
6	15	F	OD	−0.1	–	Small, elevated	EBS [−1.13]	Calcified ONHD
			OS	0.0	–	Small	Full [−1.77]	Calcified ONHD
7	18	M	OD	0.0	–	Small, elevated	EBS [−4.09]	Calcified ONHD
			OS	0.0	–	Small, elevated	EBS [−2.73]	Calcified ONHD
8	40	F	OD	2.3*	–	Small	EBS, nasal [GVF]	Normal
			OS	0.2	–	Small, elevated	Gen depression [−6.53]	Calcified ONHD
9	53	F	OD	0.0	–	Small	Full [−0.93]	Not done
			OS	0.0	Yes	Elevated, exposed ONHD	Severely constricted [−30.12]	Not done
10	61	F	OD	0.3	–	Small, elevated, exposed ONHD	Constricted, nasal [GVF]	Not done
			OS	0.3	Yes**	Elevated, exposed ONHD	Constricted, nasal [GVF]	Not done

F = female; M = male; VA = visual acuity (logMAR); OD = right eye; OS = left eye; RAPD = relative afferent pupillary defect; – = not detected; MD = mean deviation (decibels) from 24-2 Humphrey visual field; Central = central defect; EBS = enlarged blind spot; Full = full field; Gen depression = generalized depression; GVF = Goldmann visual field; Inf>sup arcuate = inferior greater that superior arcuate defects; Nasal = nasal defect;

*VA was decreased OD due to keratoconus;

**0.3 log unit RAPD.