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Fundus Autofluorescence, Optical Coherence Tomography, and Electrophoretinogram Findings in Choroidal Sclerosis

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Abstract

Purpose—To describe fundus autofluorescence, optical coherence tomography, and electrophoretinogram findings in choroidal sclerosis.

Methods—Retrospective case series. Eight eyes of four patients with choroidal sclerosis were evaluated with fundus autofluorescence, optical coherence tomography, and electrophoretinogram testing.

Results—In all eight eyes, fundus autofluorescence imaging revealed hypofluorescent placoid lesions corresponding to areas of chorioretinal atrophy seen on stereo biomicroscopy. Prominent hyperfluorescent linear markings underlying regions of atrophic disease were observed in all eyes, likely representative of normal choroidal vessel autofluorescence. In two eyes, fundus autofluorescence revealed punctate hypofluorescent lesions in the fovea that were not visualized on biomicroscopy. In one eye, fundus autofluorescence identified a central island of preserved retinal pigment epithelium that was not appreciated on ophthalmoscopic examination. Optical coherence imaging was significant for loss of choroidal fine tubular structures, retinal pigment epithelium, and outer nuclear layer in regions of chorioretinal atrophy. Full-field electrophoretinogram testing demonstrated generalized rod-cone dysfunction in all patients with a lower b- to a- wave ratio in two patients.

Conclusion—Fundus autofluorescence and optical coherence tomography are noninvasive diagnostic adjunct that can aid in the diagnosis of choroidal sclerosis. Fundus autofluorescence may be a more sensitive marker of disease extent and progression than clinical exam alone. Electrophoretinogram testing can result in an electronegative maximal response.

Keywords

choroidal sclerosis; choroidal atrophy; fundus autofluorescence; optical coherence tomography; electronegative; electrophoretinogram; retina

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INTRODUCTION

Choroidal sclerosis is a term used to describe a group of genetically heterogeneous retinal dystrophies that share a common funduscopy phenotype of scalloped, well-circumscribed, and confluent chorioretinal atrophic lesions that extend beyond the macula. The term choroidal sclerosis was initially coined because of the sclerotic appearance of choroidal vessels which frequently appear white or light-yellow^{1–5}. However, histologic studies have demonstrated the term to be a misnomer as the principle abnormality is the loss of choriocapillaris, retinal pigment epithelium (RPE), and the outer retina in the absence of abnormal sclerotic changes in the choroidal vasculature^{6–7}. Three forms of choroidal sclerosis have been identified, reflecting its geographic predilection: central areolar, peripapillary, and diffuse choroidal atrophy. Fluorescein angiography characteristics of choroidal sclerosis are well-described with window defects from RPE atrophy, loss of choriocapillaris, and increased visualization of medium and large choroidal vessels^{8–10}.

Fundus autofluorescence (FAF) and optical coherence tomography (OCT) have become powerful imaging modalities in evaluating the photoreceptor/RPE-complex¹¹. To date, imaging characteristics have not been reported for diffuse choroidal sclerosis. The purpose of this case series is to describe FAF, OCT, and electroretinogram (ERG) findings in diffuse choroidal sclerosis to better define the phenotype.

METHODS

Patients diagnosed with choroidal sclerosis were identified retrospectively from March 2006 to December 2008 at the Edward S. Harkness Eye Institute of Columbia University Medical Center (New York, NY). Four patients were identified and medical records were reviewed. Data abstracted included patient demographics, ocular history, and best-corrected Snellen visual acuity. Color photographs, red-free photographs, fluorescein angiography, electroretinogram (ERG), OCT, and FAF studies were reviewed.

FAF imaging was performed with a confocal scanning laser ophthalmoscope (cSLO, Heidelberg Retina Angiograph 2; Heidelberg Engineering, Dossenheim, Germany) after pupil dilation with topical tropicamide and phenylephrine. FAF imaging was performed using a 30° field of view at a resolution of 1536 × 1536 pixels. An optically pumped solid-state laser (488 nm) was used for excitation and a 500 nm barrier filter was used to modulate the blue argon excitation light. Standard procedure was followed for the acquisition of FAF images, including focus of the retinal image in the infrared reflection mode at 820 nm, sensitivity adjustment at 488 nm, and acquisition of 9 single 30° × 30° FAF images encompassing the entire macular area with at least a portion of the optic disc. The 9 single images were computationally averaged to produce a single frame with improved signal-to-noise ratio.

Spectral-domain optical coherence tomography (SD-OCT, Cirrus™ HD OCT, Carl Zeiss, Dublin, CA, USA) was performed on two patients bilaterally. The acquisition protocol consisted of a five-line raster scan and a macular cube 512 × 128 scan pattern in which a 6 × 6 mm region of the retina was scanned (a total of 65,536 sampled points) within a scan time of 2.4 seconds. After image acquisition, those with a signal strength of ≤8 were excluded. The thickness of the parafoveal outer nuclear layer was also recorded. Horizontal scans through the macular area were repeated three times and images were registered and averaged.

Ganzfeld full-field ERGs (Diagnosys LLC, Lowell, USA) were recorded from both eyes with DTL electrodes according to the International Society for Clinical Electrophysiology of Vision (ISCEV) standard (see www.iscev.org), in both scotopic and photopic states to assess retinal function.

The study adhered to the tenets of the Declaration of Helsinki and received approval by the Institutional Review Board (Protocol # AAAD9323) of New York-Presbyterian Hospital (New York, NY). Medical charts were reviewed in compliance with the Health Insurance Portability and Accountability Act.

RESULTS

Case 1

A 78-year-old man with a past medical history of hypertension and aortic valve replacement was referred for poor visual acuity for the last 15 years. Family history was significant for blindness of unclear etiology in patient's mother with onset at age 38. Best-corrected visual acuity measured 20/30 in both eyes. The patient was mildly myopic with -2.00 spherical correction in both eyes. Anterior segment examination was significant for posterior chamber intraocular lenses in both eyes. Ophthalmoscopic examination was notable for dense atrophic lesions in the peripapillary and nasal regions and atrophy along the arcades. White and orange choroidal vessels were visualized within the regions of atrophy. No optic disc pallor, bony spicules, or pigmentary mottle was noted. The fovea appeared intact by biomicroscopy (Figure 1 A–B). FAF imaging identified additional RPE defects with punctate lesions in the fovea of both eyes, which were not readily apparent by ophthalmoscopic examination. An annular pattern of speckled atrophy was noted along the arcades in addition to sharply demarcated areas of RPE atrophy along the arcades, peripapillary regions, and nasal to the optic nerve. Hyperfluorescent linear markings were visible within the dense atrophic regions on FAF (Figure 1 C–D), likely representative of choroidal vessel autofluorescence. Optical coherence tomography revealed preservation of the foveal structures in both eyes (Figure 1 E–F). Full field ERG showed generalized rod-cone dysfunction with an electronegative maximal response (Table 1, Figure 1G). Ornithine levels were within normal limits.

Case 2

A 53-year-old man with no past medical history was evaluated for decreased visual acuity since his teenage years. Family history was significant for poor visual acuity in his sister (Case 3) and retinal detachment and age-related macular degeneration in his mother. Best-corrected visual acuity measured 20/60 in the right eye and 20/80 in the left eye. The patient was moderately myopic with spherical correction of -4.00 OD and -3.50 OS. Anterior segment examination was significant for mild nuclear sclerosis in both eyes. Biomicroscopy revealed confluent atrophy in the macula with a central island of preserved RPE in the right eye, multiple mid-peripheral scalloped atrophic lesions, and peripapillary sparing. White and orange choroidal vessels were visualized (Figure 2 A–B) along with scattered pigment clumping concentrated in the mid-periphery. FAF revealed similar finding but also identified a central island of sparing in the left eye, which was not readily apparent by biomicroscopy. Large hyperfluorescent linear markings were noted within the placoid hypofluorescent atrophic lesions (Figure 2 C–D). Optical coherence tomography revealed regions of chorioretinal atrophy in both eyes (Figure 2 E–F). Full field ERG demonstrated generalized rod-cone dysfunction with an electronegative maximal response (Table 1). Ornithine levels were measured and within normal limits.

Case 3

A 61-year-old woman with a past medical history of hypertension was referred for slowly progressive vision loss starting at age 21. Family history was significant for poor visual acuity in her brother (Case 2) and retinal detachment and age-related macular degeneration in her mother. Best-corrected visual acuity measured CF in the right eye and 20/50 in the left eye. The patient was moderately myopic with -4.00 spherical correction in both eyes. Anterior segment examination was significant for mild nuclear sclerosis in both eyes. Ophthalmoscopic

examination demonstrated scalloped atrophic lesions in the mid-peripheral and confluent atrophy in the macula and peripapillary regions of both eyes. Orange and white choroidal vessels were visible within atrophic regions. Pigment clumping was noted in the mid-periphery with a few lesions within the arcades (Figure 3 A–B). FAF revealed extensive, dense, scalloped RPE defects involving the fovea in both eyes (Figure 3 C–D). Hyperfluorescent linear markings were easily visible in the dense placoid regions of atrophy. Optical coherence tomography identified preservation of the foveal structure in the right eye with loss of retinal pigment epithelium, choriocapillaris, and outer nuclear layer in the left eye (Figure 3 E–F). Full field ERG showed severe generalized rod-cone dysfunction with attenuated amplitudes (Table 1). Ornithine levels were within normal limits.

Case 4

A 69-year-old woman complained of progressively worsening of peripheral vision in both eyes for the past three years. Past ocular history was significant for cataract extraction with posterior chamber intraocular lens placement two year prior in the right eye. On examination, vision was best corrected to 20/25 in the right eye and 20/20 in the left eye. The patient was mildly hyperopic with spherical correction of +1.00 OD and +1.25 OS. Anterior segment was unremarkable aside from a well-positioned posterior chamber intraocular lens in the right eye. Intraocular tensions measured 17 mmHg in the right eye and 19 mmHg in the left eye. Dilated ophthalmoscopic examination featured scalloped atrophy with rare intraretinal pigment migration in the peripheral fundus (Figure 4 A–B). White and orange choroidal vessels were visualized within the regions of atrophy. Autofluorescent imaging (Figure 4 C–D) confirmed the peripheral scalloped atrophy with good foveal preservation. Hyperfluorescent linear markings were visible in the regions of atrophy. OCT (Figure 4 E–F) demonstrated preservation of the foveal structures in both eyes. Full field electroretinogram was consistent with generalized rod-cone dystrophy. Ornithine levels were assessed previously and results were within normal limits.

DISCUSSION

Choroidal sclerosis represents a group of retina dystrophies that share a common phenotype of scalloped, well-circumscribed, and confluent chorioretinal atrophic lesions that extend beyond the macula. This case series identifies FAF and OCT imaging characteristics and ERG findings in choroidal sclerosis. In all cases, FAF identified areas of disease with loss of choriocapillaris fenestration and overlying RPE. These regions manifested as placoid hypofluorescent lesions with hyperfluorescent linear markings, which may represent visualization of normal choroidal autofluorescence within the regions of overlying atrophy. FAF also detected RPE changes that were not evident on ophthalmoscopic examination. In case 1, FAF identified punctate lesions in the fovea of both eyes that were not visible on biomicroscopy, demonstrating more widespread involvement of the RPE than originally thought. In case 2, FAF revealed a central island of RPE sparing in the left eye that was not appreciated on biomicroscopy, signaling less severe disease progression. OCT findings demonstrated increased signals reflected from choroidal layers, loss of fine tubular structures of the choriocapillaris, and outer nuclear layer loss in regions of chorioretinal atrophy. ERG testing was significant for generalized rod-cone dysfunction in all cases and an electronegative maximal response in cases 1 and 2. FAF, OCT, and ERG testing may be useful adjuncts in diagnosis and FAF may be a more sensitive marker of disease extent than clinical exam alone.

FAF imaging relies on the stimulated emission of light from molecules, chiefly lipofuscin, in the RPE and provides indirect information on the level of metabolic activity of the RPE¹³. Hyperautofluorescence is thought to correspond to accumulation of lipofuscin in the RPE or subretinal space. Excessive lipofuscin accumulation represents a common pathogenetic

pathway in various degenerative and monogenetic retinal diseases and is believed to precede photoreceptor degeneration^{14–15}. A prominent component of lipofuscin is a pyridinium bisretinoid, A2E (*N*-retinyl-*N*-retinylidene ethanolamine), which possesses detergent properties and may interfere with normal RPE lysosomal function¹⁶. Longitudinal observations have suggested that increased accumulation of autofluorescent material may occur before cell loss¹⁷. Hypoautofluorescence is thought to correspond to areas of decreased metabolism resulting from photoreceptor and/or RPE atrophy and has been used as a marker for the integrity of the RPE/photoreceptor complex^{18–20}. In the choroid, autofluorescence of elastin and red and white blood cells results in hyperfluorescent vascular markings that are typically blocked in healthy eyes by the overlying choriocapillaris and RPE. However, the choroidal vasculature may be visualized on FAF in diseases in which the overlying layers become atrophic¹¹.

The choroidal sclerosis phenotype includes genetically heterogeneous diagnoses such as choroideremia, gyrate atrophy, *PRPH2*/rds-associated retinal degeneration²¹, *ABCA4*-disease²², *RLBP1*-associated retinitis punctata albescens^{23–24}, cone rod dystrophies associated to *CERKL* mutations²⁵, pericentral retinal degenerations²⁶, and late-onset retinal degeneration²⁷. Identification of the choroidal sclerosis funduscopy pattern should guide molecular clarification of the suspected genes and help prioritize future gene screening. A precise molecular diagnosis will facilitate rapid identification of appropriate candidates for targeted therapies.

FAF, OCT, and ERG testing in conjunction with clinical examination and fluorescein angiography are helpful in the diagnosis and evaluation of choroidal sclerosis. FAF may be particularly valuable in follow-up of patients as small lesions, which may not be detectable by clinical examination, may be easily and non-invasively identified and allow for close monitoring of disease progression.

SUMMARY STATEMENT

Fundus autofluorescence and optical coherence tomography can aid in the diagnosis of choroidal sclerosis and may be a more sensitive marker of disease extent and progression than clinical exam alone. Electroretinogram testing can sometimes demonstrate a lower b- to a- wave ratio.

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Figure 1.

Case 1 Imaging Studies. Color fundus photographs (A–B) reveal chorioretinal atrophy in the juxtapapillary and peripapillary regions that extend along the arcades. Orange and white choroidal vessels are visible in regions of atrophy. No optic disc pallor, bony spicules, or pigmentary mottling is noted. Fundus autofluorescence (C–D) reveals more extensive retinal pigment epithelium defects with speckled hypofluorescent foveal lesions in both eyes that were not visualized on clinical examination. An annular pattern of retinal pigment epithelium defects is noted with sharply demarcated lesions in the peripapillary regions, along the arcades, and nasal to the optic disc. Hyperfluorescent linear markings are noted within the placoid regions of atrophy, possibly representing visualization of large choroidal vessels within the regions of overlying retinal pigment epithelium atrophy. Optical coherence tomography (E–F) reveals preserved foveal architecture in both eyes with thinning and regions of chorioretinal atrophy in the perifoveal region. Electroretinogram (G) demonstrates electronegative maximal dark-adapted responses with low b-a wave ratios.

Figure 2.

Case 2 Imaging Studies. Color fundus photographs (A–B) demonstrate mid-peripheral scalloped lesions and confluent central atrophy. A central island of preserved retinal pigment epithelium is noted in the right eye. White and orange markings representing the choroidal vasculature are visible within the atrophic lesions. Pigmentary clumping is concentrated in the mid-periphery. Fundus autofluorescence (C–D) identifies a central island of retinal pigment epithelium sparing in the left eye, which was not appreciated with biomicroscopy. Hyperfluorescent linear markings are visible within the atrophic hypofluorescent placoid regions. Optical coherence tomography (E–F) reveals regions of atrophy of the retinal pigment epithelium, loss of fine fenestrated choroidal vasculature, and outer nuclear layer in both eyes. Increased visualization of choroidal vessels is seen in regions devoid of retinal pigment epithelium.

Figure 3.

Case 3 Imaging Studies. Color fundus photographs (A–B) demonstrate scalloped lesions in the mid-periphery and confluent atrophy of the macula and peripapillary regions. Orange and white choroidal vessels are visible in areas of atrophy. Pigmentary clumping is predominantly in the mid-periphery. Fundus autofluorescence (C–D) reveals extensive, scalloped retinal pigment epithelium defects involving the fovea in both eyes. Fundus autofluorescence of the peripapillary region of the left eye (D) demonstrates changes in backscattering in areas of chorioretinal atrophy with hyperfluorescent linear markings likely representative of normal choroidal autofluorescence unmasked by overlying atrophy. Optical coherence tomography (E–F) reveals loss of foveal retinal pigment epithelium, loss of fine fenestrated choriocapillaris, and outer nuclear layer in the right eye with relative preservation of the foveal structures in the left eye. Increased visualization of choroidal vessels is seen in regions devoid of retinal pigment epithelium.

Figure 4.

Case 4 Imaging Studies. Color fundus photographs (A–B) demonstrate mid-peripheral scalloped lesions in both eyes with sclerotic appearing white and orange choroidal vessels most prominently noted within the atrophic lesions. There is rare intraretinal pigment migration peripherally. Fundus autofluorescence (C–D) confirms the scalloped atrophy in the periphery with preservation of the fovea in both eyes. Hyperfluorescent linear markings are visible within the atrophic regions. Optical coherence tomography (E–F) reveals preservation of the foveal structures in both eyes.

Table 1

Full-Field ERG Findings

Stimulus	OD		OS		Normal Values			
	Amplitude (mV)		Implicit Time (ms)		Amplitude (mV)		Implicit Time (ms)	
	A-Wave	B-Wave	A-Wave	B-Wave	A-Wave	B-Wave	A-Wave	B-Wave
Rod-specific								
<ems>Case 1	30			30			191 [plusmn]	123
<ems>Case 2	5			3			250 [plusmn]	143
<ems>Case 3	4			5			241 [plusmn]	117
<ems>Case 4	6			6			198 [plusmn]	137
Maximal ERG								
<ems>Case 1	42	30		36	159 [plusmn]	62	338 [plusmn]	209
<ems>Case 2	15	6	36	5	180 [plusmn]	106	333 [plusmn]	201
<ems>Case 3	10	11	12	11	180 [plusmn]	106	333 [plusmn]	201
<ems>Case 4	14	33	18	31	159 [plusmn]	62	338 [plusmn]	209
Cone single flash								
<ems>Case 1	25	34		10	144 [plusmn]	66	29 [plusmn]	2
<ems>Case 2	12	41		17	129 [plusmn]	47	28 [plusmn]	2
<ems>Case 3	12	49		9	129 [plusmn]	47	28 [plusmn]	2
<ems>Case 4	13	40		18	144 [plusmn]	66	29 [plusmn]	2
30-Hz flicker								
<ems>Case 1	21	36		24	108 [plusmn]	40	24 [plusmn]	3
<ems>Case 2	12	42		25	107 [plusmn]	33	24 [plusmn]	2
<ems>Case 3	9	42		11	107 [plusmn]	33	24 [plusmn]	2
<ems>Case 4	11	34		16	108 [plusmn]	40	24 [plusmn]	3

All cases showed generalized rod-cone dysfunction and cases 1 and 2 showed an electronegative maximal response. Case 3 showed severe dysfunction with attenuated amplitudes. OD, right eye; OS, left eye.