OPEN

ATYPICAL MACULOPATHY IN A PATIENT WITH LIGHT CHAIN DEPOSITION DISEASE MIMICKING ADVANCED GEOGRAPHIC ATROPHY

Courtney Crawford, MD,* Lauren J. Oshry, MD,† Elias Reichel, MD*

Purpose: To report a previously unreported presentation of advanced geographic atrophy of the macula mimicking nonneovascular (dry) age-related macular degeneration in a patient with light chain deposition disease.

Methods: Ocular examination included dilated fundus examination, fundus autofluorescence, full-field electroretinography, and spectral domain optical coherence tomography.

Patients: Single-patient case report.

Results: Dilated fundus examination demonstrated diffuse loss of the retinal pigment epithelium in a geographic atrophy pattern in the macula and drusenlike deposits localized to the outer retina and retinal pigment epithelium. There were no signs of choroidal neovascularization or retinal pigment epithelium detachments. Fundus autofluorescence demonstrated wide areas of retinal pigment epithelium loss. Full-field electroretinography was normal. Spectral domain optical coherence tomography displayed atrophy of the outer retinal layers.

Discussion: This is the first documented case of drusenlike deposits and maculopathy in a patient with light chain deposition disease that mimics advanced geographic atrophy that is typically observed in nonneovascular age-related macular degeneration. Physicians should be aware of the macular changes that can be associated with light chain deposition disease, and patients with light chain deposition disease should be regularly evaluated for associated macular disease.

RETINAL CASES & BRIEF REPORTS 11:83–85, 2017

*From the *New England Eye Center, Tufts University School of Medicine, Boston, Massachusetts; and †Boston University School of Medicine, Boston, Massachusetts.*

Light chain deposition disease (LCDD) is a rare disease characterized by the deposition of nonamyloid immunoglobulin light chains in multiple organs, most commonly in the kidney.[1] (Figure 1, A–D) An overproduction of kappa light chains is responsible for the disease, occurring more commonly in women than men.[1] Late-stage progression of renal complications includes hypertension, nephrotic syndrome and end-stage renal disease.[2] The incidence of ocular disease with LCDD is rare.[3,4] This report illustrates a previously unreported presentation of advanced geographic atrophy of the macula mimicking nonneovascular (dry) age-related macular degeneration in a patient with LCDD.

A 61-year-old woman presented with progressive decreased central vision and diminished night vision over a 10-year period. She was referred for evaluation of an atypical form of age-related macular degeneration. She had been diagnosed and treated for kidney-biopsy–proven LCDD in 2002. Best-corrected vision at presentation was 20/50 in the right eye and 20/200 in the left eye. Dilated fundus examination demonstrated diffuse loss of the retinal pigment epithelium (RPE) in a geographic atrophy pattern in the macula

Supported in part by a Research to Prevent Blindness Unrestricted grant to the New England Eye Center/Department of Ophthalmology, Tufts University School of Medicine.

None of the authors have any conflicting interests to disclose.

Reprint requests: Courtney Crawford, MD, New England Eye Center, Tufts Medical Center, 800 Washington Street, Boston, MA 02111; e-mail: courtneymcrawford@gmail.com

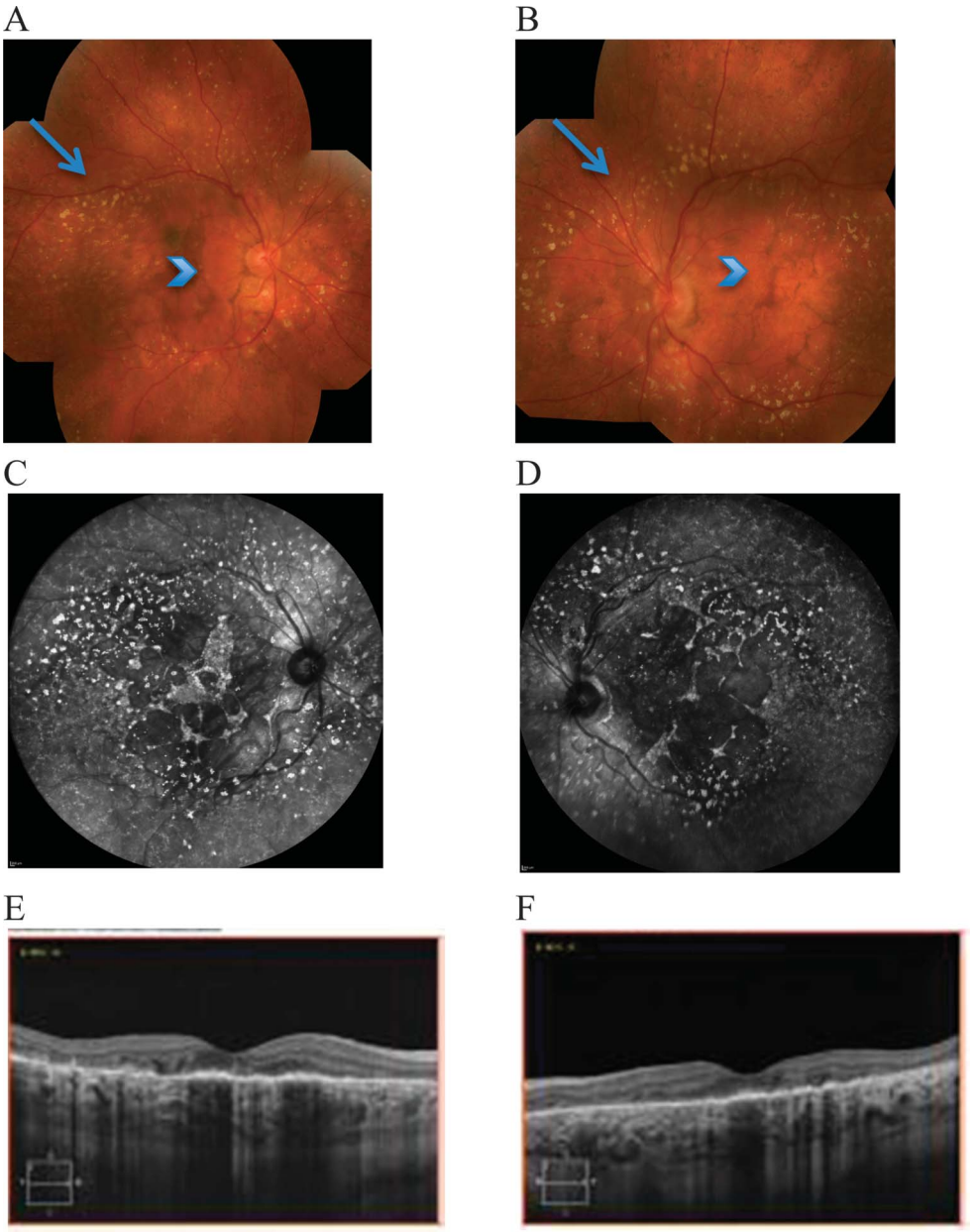

Fig. 1. Right eye (**A**) and left eye (**B**), color fundus photograph montage depicting drusen (arrows) circumferential to the macular atrophy and retinal pigment epithelium defects (arrowheads) within the macula consistent with geographic atrophy. Right eye (**C**) and left eye (**D**), fundus autofluorescence highlights drusen and areas of retinal pigment epithelium atrophy. **E.** Right eye, ocular coherence tomography depicting severe diffuse loss of the outer retina layer and retinal pigment epithelium. **F.** Left eye, ocular coherence tomography depicting severe diffuse loss of the outer retina layer and retinal pigment epithelium.

and drusenlike deposits localized to the outer retina and RPE (Figure 1, A and B). There were no signs of choroidal neovascularization or RPE detachments. Fundus autofluorescence demonstrated wide areas of RPE loss (Figure 1, C and D). Full-field electroretinography was normal. Spectral domain optical coherence tomography displayed atrophy of the outer retinal layers (Figure 1, E and F).

In this patient with LCDD and maculopathy, we believe that light-chain deposits can appear as drusen in the macula and are associated with progressive damage to the RPE and outer layers of the retina similar to what can be observed in nonneovascular age-related macular degeneration. It has been shown that the molecular composition of drusen in age-related macular degeneration can include immunoglobulin light-chains suggesting a commonality to what was observed in this patient.[5] Alternative diagnosis including pattern dystrophy and Stargardt macular dystrophy was considered but ruled out because of the lack of typical examination findings to include lipofuscin deposits and the patient's late age of onset. A postmortem histopathological analysis, of a patient with LCDD and RPE detachments, showed extensive deposition of kappa light chains in the RPE and choroid.[4] Only one other case report describes vision loss, from RPE detachments, in three

patients with LCDD; serous retinal detachments were reported but atypical drusen representing light-chain deposits were not described in any of these patients.[3,4] It is possible that our patient experienced serous retinal detachments before her presentation; however, no sign of subretinal fluid or choroidal neovascular membranes were present on examination.

This is the first documented case of drusenlike deposits and maculopathy in a patient with LCDD that mimics advanced geographic atrophy that is typically observed in nonneovascular age-related macular degeneration. Physicians should be aware of the macular changes that can be associated with LCDD, and patients with LCDD should be regularly evaluated for associated macular disease.

Key words: macular degeneration, light chain deposition disease, geographic atrophy.

References

1. Ronco P, Plaisier E, Mougenot B, Aucouturier P. Immunoglobulin light (heavy)-chain deposition disease: from molecular medicine to pathophysiology-driven therapy. Clin J Am Soc Nephrol 2006;1:1342–1350.
2. Pozzi C, D'Amico M, Fogazzi GB, et al. Light chain deposition disease with renal involvement: clinical characteristics and prognostic factors. Am J Kidney Dis 2003;42:1154–1163.
3. Daicker BC, Mihatsch MJ, Strøm EH, et al. Ocular pathology in light chain deposition disease. Eur J Ophthalmol 1995;5: 75–81.
4. Spielberg LH, Heckenlively JR, Leys AM. Retinal pigment epithelial detachments and tears, and progressive retinal degeneration in light chain deposition disease. Br J Ophthalmol 2013;97:1218–1219.
5. Mullins RF, Russell SR, Anderson DH, Hageman GS. Drusen associated with aging and age-related macular degeneration contain proteins common to extracellular deposits associated with atherosclerosis, elastosis, amyloidosis, and dense deposit disease. FASEB J 2000;14:835–846.