

Case Report

A Case of Extensive Macular Atrophy with Pseudodrusen-Like Appearance

Shinji Makino* and Hironobu Tampo

Department of Ophthalmology, Jichi Medical University, Japan

*Corresponding author

Shinji Makino, Department of Ophthalmology, Jichi Medical University, Shimotsuke, 3311-1 Yakushiji, Tochigi 329-0498, Japan; Tel: +81-285-58-7382 ; Fax: +81-285-44-8365; E-mail: makichan@jichi.ac.jp

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Abstract

A 62-year-old woman complaining of night blindness and unclear vision in both eyes for the previous 10 years was referred to our hospital. Her best-corrected visual acuity was 0.6 in both eyes. Fundus examination showed geographic macular atrophy with increased vertical diameter in both eyes. The macular atrophy was surrounded by numerous drusen-like changes spread throughout the posterior pole and mid periphery of the retina. Visual acuity decreased to 0.3 six months after the initial visit, and 0.03 one year after the initial visit, in both eyes. Reevaluation 54 months after the initial visit revealed visual acuity of 0.02 in both eyes. On fundus examination, the atrophy had progressed with increased visibility of the choroidal blood vessels. The pseudodrusen-like changes were still visible all around the atrophy. In addition, paving-stone degeneration was detected in the temporal peripheral retina in both eyes. On optical coherence tomography, macular thickness was reduced, the photoreceptor line was undetectable, and hyperreflective punctate drusen-like substances were clearly detected in the atrophic area. From the above findings, we diagnosed extensive macular atrophy with pseudodrusen-like appearance.

Keywords

- Extensive macular atrophy with pseudodrusen-like appearance
- Macular atrophy
- Paving-stone degeneration
- Optical coherence tomography

INTRODUCTION

In 2009, Hamel et al. [1] proposed the disease “extensive macular atrophy with pseudodrusen-like appearance” (EMAP) as a new clinical entity. It is characterized by early-onset (<50 years) bilaterally symmetric macular atrophy with increased vertical axis, surrounded by numerous pseudodrusen-like deposits, with rapid involvement of the fovea and entire posterior pole. In addition, EMAP patients have presented with paving-stone degeneration predominantly in the inferior part of the peripheral retina. To our knowledge, there are two reports in the literature describing EMAP [1,2]. Furthermore, there is one report describing the optical coherence tomography (OCT) findings in EMAP patients [1]. Herein, we report a single case of EMAP.

CASE PRESENTATION

A 62-year-old woman complaining of night blindness and blurry vision in both eyes for the previous 10 years was referred to our hospital. The patient’s family and personal medical history added no significant information. Her best-corrected visual acuity was 0.6 in both eyes. On slit-lamp examination, mild cortical opacities were detected in both lenses. Ocular pressures were normal. Fundus examination showed geographic macular atrophy in both eyes (Figure 1A and B). The macular atrophy was surrounded by numerous drusen-like changes including small white punctate spots and poorly defined flat spots spread

throughout the posterior pole and mid periphery of the retina. Fluorescein angiography (FA) showed a transmission defect with granular hyperfluorescence and increased vertical diameter (Figure 2A and B). Goldmann perimetry disclosed an absolute central scotoma (20 central degrees) sparing the fovea in both eyes (Figure 3A and B). Full-field ERG showed moderately reduced amplitude in both eyes (Figure 4). On optical coherence tomography (OCT), macular thickness was reduced in both eyes. Six months after the initial visit, her visual acuity had decreased to 0.3, and one year after the initial visit, visual acuity was 0.03 in both eyes. The patient was reevaluated 54 months after the initial visit. Visual acuity had decreased to 0.02 in both eyes. On fundus examination, the atrophy had progressed with increased visibility of the choroidal blood vessels (Figure 5A and B). The pseudodrusen-like changes were still visible all around the

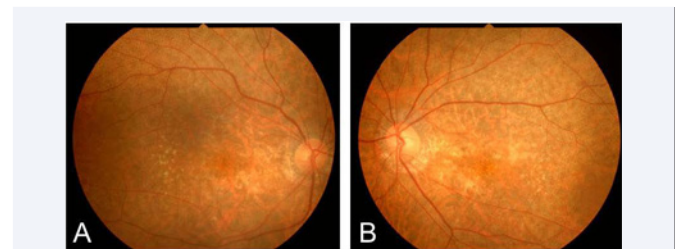


Figure 1 Right (A) and left (B) fundus photographs at the initial visit showing geographic macular atrophy.

atrophy. In addition, paving-stone degeneration was detected in the temporal peripheral retina in both eyes (Figure 6A and B). Interestingly, several types of retinal changes were detected, including paving-stone degeneration, small white punctate spots, and poorly defined flat spots (Figure 7A, B, and C, respectively). On fundus autofluorescence (FAF) imaging, the atrophy was dark and well delineated in both eyes (Figure 8A and B). On OCT (RS-3000; NIDEK, Gamagori, Japan), macular thickness was reduced to 197 μm in the right eye and 193 μm in the left eye; further, the photoreceptor line was undetectable, the choroidal signal was enhanced inside the atrophic area in both eyes (Figure 9A and B), and hyperreflective punctate drusen-like substances were clearly detected above the retinal pigment epithelium (RPE). A Heidelberg Retina Angiograph 2 (Heidelberg Engineering, Heidelberg, Germany) was used to perform FA and FAF imaging. The dark-adaptation examination was not available in this case. The patient continues to receive low vision rehabilitation.

DISCUSSION

In this case, we initially suspected the disease as cancer-associated retinopathy or bilateral diffuse uveal melanocytic proliferation [3], rather than geographic atrophy associated with age-related macular degeneration (AMD) [4], on the basis of the following findings: night blindness, rapid progression to central vision loss, and transmission defect with granular hyperfluorescence on FA. However, her blood laboratory findings including tumor markers, chest radiography, and abdominal CT were unremarkable. This patient fulfilled the inclusion criteria for EMAP: macular atrophy, drusen-like deposits in the mid periphery of the retina, and paving-stone changes in the far peripheral retina. In particular, the numerous poorly defined flat spots (Figure 7C), which looked like drusen, spread throughout the posterior pole and the mid periphery of the retina in our case were very similar to those noted in the original case description

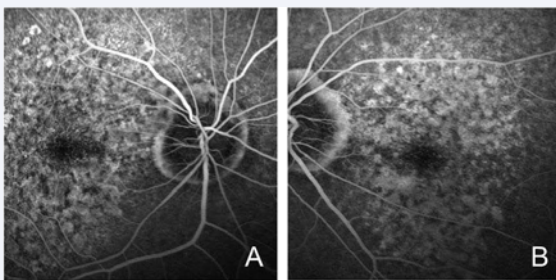


Figure 2 Right (A) and left (B) fluorescein angiography images at the first visit demonstrate a transmission defect with granular hyperfluorescence and increased vertical diameter.

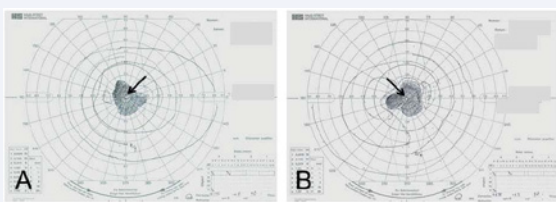


Figure 3 Right (A) and left (B) Goldmann perimetry diagrams show an absolute central scotoma (20 central degrees) sparing the fovea (arrows).

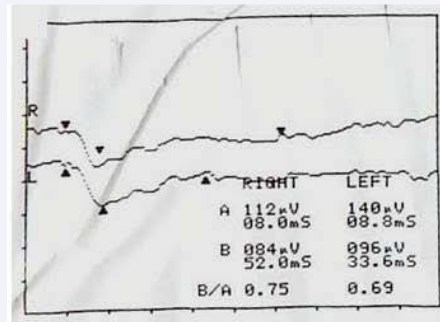


Figure 4 ERG response shows reduced amplitude in the both eyes.

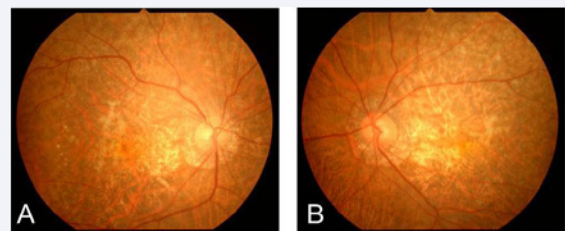


Figure 5 Right (A) and left (B) fundus photographs 54 months after the initial visit. The atrophy has progressed with increased visibility of the choroidal blood vessels.

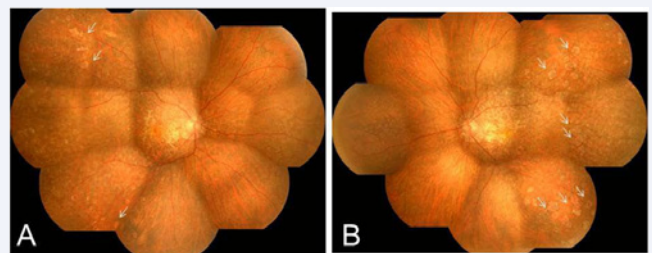


Figure 6 Right (A) and left (B) panoramic montages of fundus photographs. Paving-stone degeneration is visible in the temporal peripheral retina in both eyes (arrows).

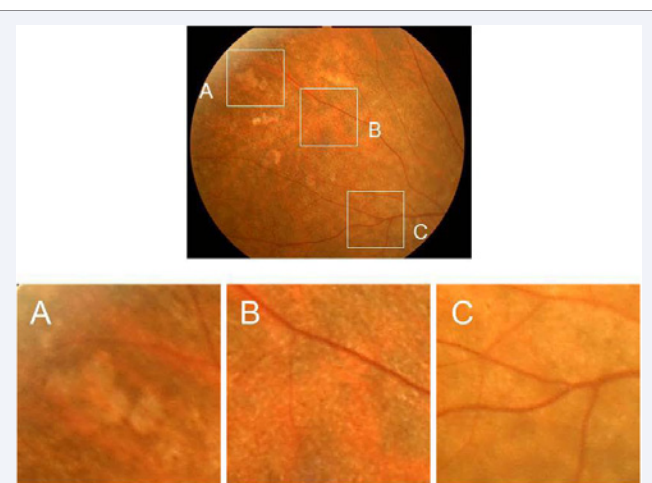


Figure 7 Magnified fundus photographs of the superotemporal peripheral retina of the right eye. A: Paving-stone degeneration. B: Small white punctate spots. C: Poorly defined flat spots.

[1]. Therefore, we diagnosed EMAP. EMAP is a recently reported rare clinical entity resembling dry AMD but with a distinct clinical appearance. Hamel et al. [1] first reported 18 patients (11 women and 7 men) with an atrophic macular disorder resembling dry AMD. The mean age was 47.5 years (range, 41–54 years) at onset and 53.5 years (range, 49–60 years) at inclusion. The most striking feature was early onset of bilaterally symmetric macular atrophy, on average before the age of 50 years, with rapid involvement of the fovea and entire posterior pole up to the temporal vascular arcades. The macular atrophy was surrounded by numerous drusen-like deposits spread throughout the posterior pole and mid periphery of the retina in all cases. The atrophic lesion was polycyclic with an oval shape and increased vertical diameter. Geographic atrophy of the RPE and choriocapillaris was bilateral in all cases and usually symmetrical. During follow-up, the atrophy generally spread rapidly into the fovea. Besides macular atrophy, all patients displayed drusen-like patterns mimicking clusters of small flat drusen. These lesions were located all around the central atrophy up to the vascular temporal arcades and in the entire mid periphery of the retina. Smith et al. [5] reported reticular macular disease including reticular pseudodrusen and basal laminar drusen. In our patient, FA findings (Figure 2A and B) were similar to those in basal laminar drusen (cuticular

drusen). Basal laminar drusen were first described by Gass et al. [6] as small round yellow lesions randomly scattered in the macula and in the mid periphery of the retina. Compared with typical AMD drusen, basal laminar drusen fluoresce during the early arteriovenous phase, exhibiting a typical “stars-in-the-sky” appearance. Vitelliform macular detachment is frequently associated with basal laminar drusen. Querques et al. [7] analyzed OCT findings in basal laminar drusen. In their report, the OCT scan showed small confluent “dome-shaped” RPE elevations. Zweifel et al. [8] described OCT findings of reticular pseudodrusen. The OCT scans showed collections of granular hyperreflective material above the RPE, in the subretinal space located primarily between the RPE and the boundary between the inner and outer segments of the photoreceptors (IS/OS boundary). In a more advanced stage, this material formed small mounds that broke through the IS/OS boundary. In our case, the location of the drusen-like substances at the posterior pole was similar to that of reticular drusen reported by Zweifel et al. [8], and mixed reticular patterns resembling basal laminar drusen and reticular pseudodrusen [5,7,8,9] were revealed on FA and OCT. There is one report describing the OCT findings in EMAP patients [1]. The OCT scan showed decreased macular thickness and an enhanced choroidal signal, and the photoreceptor line was undetectable in all eyes. On peripheral OCT scans performed in the location of the drusen-like lesions, no nodular thickening of the RPE-Bruch membrane complex was disclosed. Although peripheral OCT scans were not available in this case, hyperreflective punctate drusen-like materials were clearly detected above the RPE in the posterior pole. Hamel et al. [1] reported all EMAP patients showed paving-stone degeneration in the far periphery, predominantly in the inferior part of the peripheral retina. Querques et al. [2] reported similar findings on ultra-wide-field color imaging. In contrast, paving-stone changes were mainly located in the temporal retina, rather than the inferior peripheral retina, in their cases. In our case, paving-stone degeneration was located in the temporal peripheral retina in both eyes. O’Malley et al. [10] studied a series of 1223 eyes obtained from 614 individuals at autopsy and found paving-stone degeneration in 27% of subjects over 20 years of age. Additionally, lesion distribution according to quadrant was as follows: inferotemporal, 78.5%; superotemporal, 15.6%; superonasal, 8.6%; and inferonasal, 5.7%. Considering this prevalence and location of paving-stone degeneration, it is not clear whether a significant association exists between EMAP and paving-stone degeneration. Further study and additional cases are necessary to answer this question. EMAP should be distinguished from dry AMD and several other diseases including cone dystrophy, cone rod dystrophy, inverse retinitis pigmentosa, Sorsby fundus dystrophy, North Carolina macular dystrophy, central areolar choroidal dystrophy, late-onset macular retinal dystrophy, basal laminar drusen combined with vitelliform macular detachment, and retinal dystrophy associated with maternally inherited diabetes and deafness [1,2,11]. Further, extensive chorioretinal atrophy of the macula, with or without drusen, may be the end stage of a broad range of genetically well-defined retinal dystrophies [11]. Boon et al. [11] recommend screening for mutations of known and plausible genes. As an example, Boon et al. [12] strongly suggest that monogenic inheritance of *CFH* variants can result in basal laminar drusen in young adults, and this can progress to maculopathy and

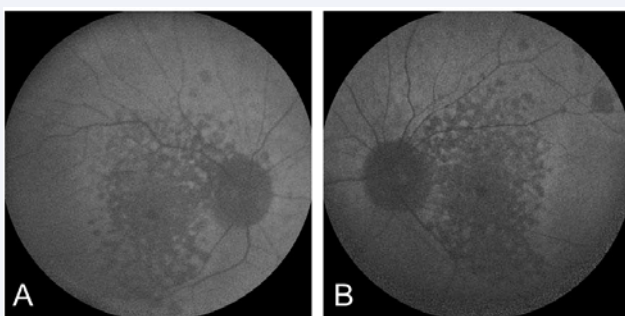


Figure 8 Right (A) and left (B) fundus autofluorescence images. The atrophy is dark and well delineated in both eyes.

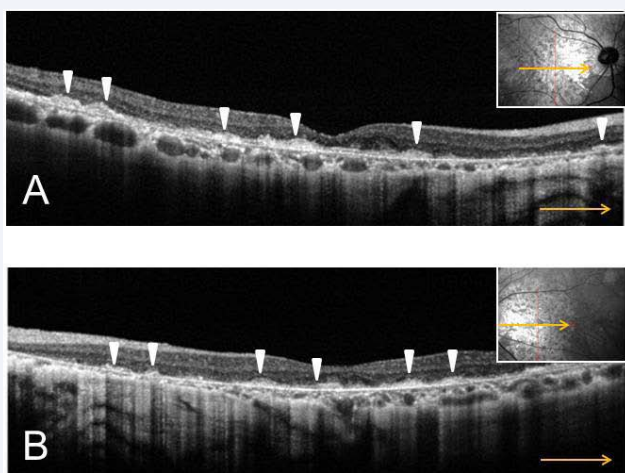


Figure 9 Right (A) and left (B) optical coherence tomography images at the posterior pole. Macular thickness is reduced, the photoreceptor line is undetectable, and the choroidal signal is enhanced. Hyperreflective punctate drusen-like materials are visible (arrowheads).

severe vision loss later in life. Our current findings were based on a single case; further, the dark-adaptation examination, red-free or blue-light fundus pictures [5], and peripheral OCT scans were not available. Therefore, the pathological layers of the retina detected as the small white punctate spots and the poorly defined flat spots were not clearly identified. Additional cases and genetic screening are needed for definitive characterization of EMAP.

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