

Fundus Autofluorescence Imaging and Optical Coherence Tomography Analysis in a Patient with Stargard Disease

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Abstract: We present a case of Stargardt disease in a 66-year-old man. Fundus examination revealed atrophic macula in both eyes. Fundus autofluorescence (FAF) imaging showed clearly defined hypo-autofluorescent lesions corresponding to the atrophic lesion. In addition, numerous hyper-autofluorescent dots corresponding to the retinal flecks were observed around the atrophic area. Near-infrared FAF imaging showed central hyper-autofluorescent lesions corresponding to the hypo-autofluorescent dark area examined by FAF. Optical coherence tomography (OCT) revealed disruption of the photoreceptor inner segment/outer segment interface. Some retinal flecks were detected on OCT. Retinal flecks were more easily defined on FAF images than on color photographs. FAF was useful in visualizing retinal flecks even though the end-stage Stargardt disease.

Keywords: Stargardt disease, retinal flecks, fundus autofluorescence imaging, optical coherence tomography.

INTRODUCTION

Stargardt disease is a typical inherited macular dystrophy that appears mostly in young individuals[1]. On fundus examination, Stargardt disease is characterized by a progressive atrophic macular area surrounded by few or numerous ill-defined yellowish flecks[1]. Generally, retinal flecks vary in size and shape. They have a yellow-whitish color and are well defined at the early stages of the disease process. They often become hazy, grey, ill-defined and barely detectable on fundus examination[2, 3]. However, the flecks are clearly evident on fundus autofluorescence (FAF) as hyper-autofluorescent or sometimes hypo-autofluorescent areas in the later stages of the disease[2-6]. We describe FAF and optical

coherence tomography (OCT) findings in a patient with Stargardt disease.

CASE REPORT

A 68-year-old Japanese man was referred to our clinic for a 10-year history of bilateral unclear vision and one-year history of bilateral central scotoma. His medical history was unremarkable, and there was no family history of ocular disease. On ophthalmic examination, his best corrected visual acuity was 0.05 in the right eye and 0.09 in the left eye, both anterior segments were normal, and the ocular pressures were normal. Ophthalmoscopy of both eyes revealed atrophic macula with increased visibility of the choroidal blood vessels (Figures 1A and 1B arrows).

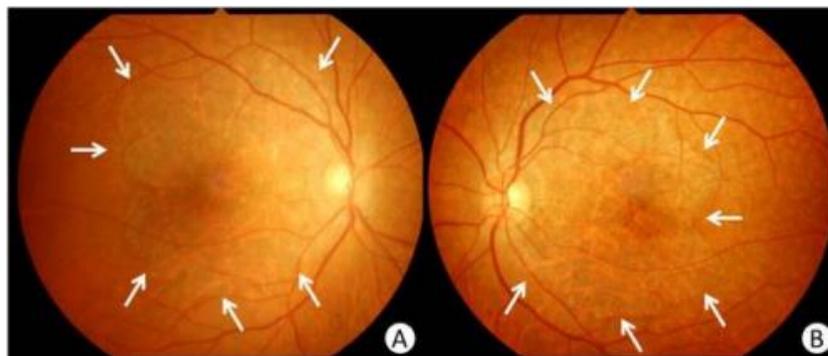


Fig.1 Fundus photographs of the (A) right and (B) left eyes.

Note macular atrophy with increased visibility of the choroidal blood vessels (arrows). FAF (Heidelberg Retina Angiograph 2, Heidelberg

Engineering, Heidelberg, Germany) imaging showed clearly defined hypo-autofluorescent lesions corresponding to the atrophic lesion in both eyes

(Figures 2A and 3A). In addition, numerous hyper-autofluorescent lesions corresponding to the retinal flecks were observed around the atrophic area. Near-infrared FAF (NIR-FAF) imaging showed central hyper-autofluorescent lesions corresponding to the hypo-autofluorescent dark area examined by conventional FAF (Figures 2B and 3B). Retinal flecks

were more easily defined on FAF images than on color photographs and NIR-FAF.OCT (DRI OCT-1 Atlantis; TOPCON, Japan) revealed the disruption of the inner segment/outer segment (IS/OS) interface. Some retinal flecks were detected above the retinal pigment epithelium (RPE) (Figures 2C and 3C, arrowheads).

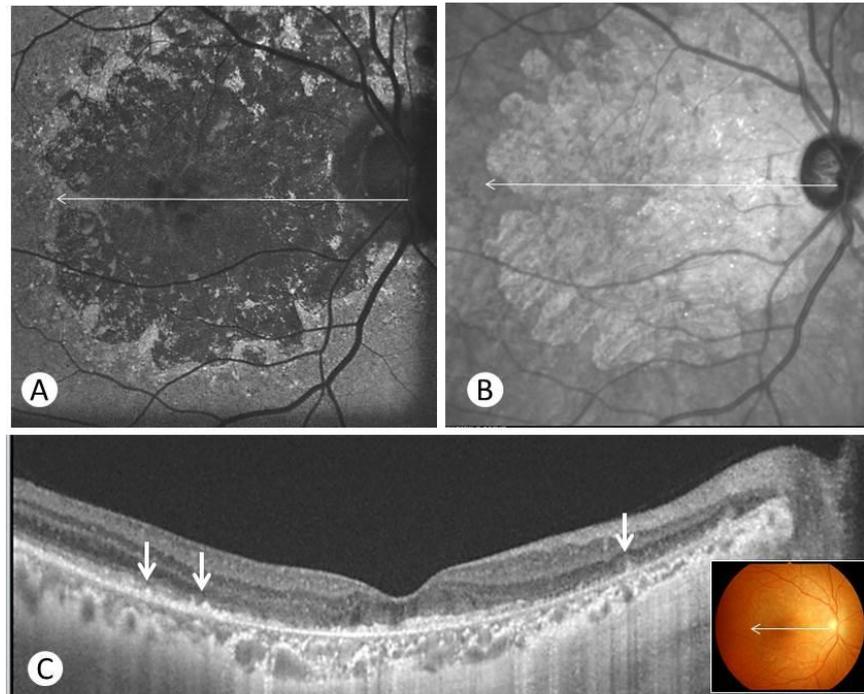


Fig.2 FAF (A), NIR-FAF (B) and OCT (C) imaging of the right eye.

FAF shows clearly defined hypo-autofluorescent dark area surrounded by flecks. NIR-

FAF shows hyper-autofluorescence. Flecks were detected above the RPE (arrowheads).

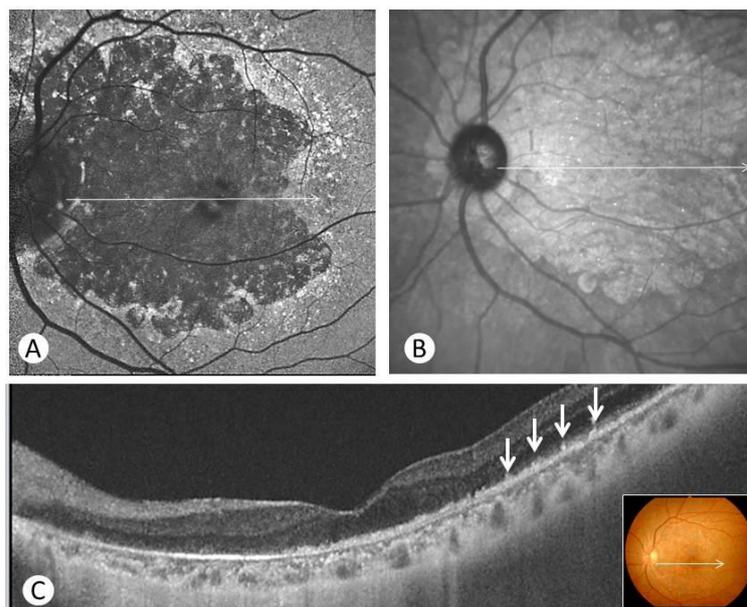


Fig.3 FAF (A), NIR-FAF (B) and OCT (C) imaging of the left eye.

FAF shows clearly defined hypo-autofluorescent dark area surrounded by flecks. NIR-FAF Shows hyper-autofluorescence. Flecks were detected above the RPE (arrowheads).

Fluorescein angiography revealed hyperfluorescence, and indocyanine green angiography showed hypofluorescence within the lesion. Visual field testing by Goldmannperimetry showed absolute central scotoma (15 central degrees) in both eyes. The standard electroretinogram including flicker, flash, rod response and cone response moderately reduced amplitude in both eyes. Multifocal electroretinograms showed reduced amplitude. Based on these collective findings, we diagnosed our patient with end-stage of Stargardt disease. The visual findings did not change during the 6-month follow-up period.

DISCUSSION

In this case, we initially suspected the disease as peripapillary acute zonal occult outer retinopathy with drusen-like materials[7, 8] or geographic atrophy associated with age-related macular degeneration[9, 10]on the basis of the ophthalmoscopic findings. However, the numerous retinal flecks were detected on FAF. Therefore, we diagnosed Stargardt disease.

Typical morphologic features of the early-stage Stargardt disease are the yellowish-white well-defined deposits, the so-called retinal “flecks.” As the central atrophy progresses, it becomes increasingly difficult to distinguish the flecks on fundus examination[2]. Furthermore, the flecks, which are clearly revealed by FAF as hyper-autofluorescent during the early stages of the disease, appear hypo-autofluorescent in FAF at the end-stage disease, suggesting a turn over from material accumulation until complete resorption and cell death[2-6].In this present case, retinal flecks were more easily defined on FAF images than on color photographs.

On OCT, the flecked areas corresponded to hyper reflective lesions at the level of the RPE associated with a dislocation or disruption of the IS/OS line [2-6]. Voigt et al. [2] proposed a classification for retinal flecks in Stargardt disease based on OCT findings: the classifications comprise 5 distinct types of lesion in relation to their localization in the outer retinal layers. According to their report, Group A lesions were limited to the OS of the photoreceptors, the RPE, and the RPE/Bruch membrane complex. Group B showed a protrusion of the hyper-reflective material through the interface of IS/OS of the photoreceptors up to the external limiting membrane. A further protrusion of the hyperreflective material into the outer nuclear layer was seen in group C lesions. Group D lesions were characterized by an accumulation of the hyper-reflective material limited to the outer nuclear layer.

Type E lesions can be described as drusen-like retinal pigment detachments. In this present case, retinal flecks were detected as drusen-like lesions.

Conventional FAF can visualize lipofuscin in the RPE[5]. In contrast,NIR-FAF appearsto correspond to melaninrather than lipofuscin[5]. Generally, retinal flecks are detected as hyper-autofluorescent on FAF, and as hypo-autofluorescent on NIR-FAF. Although our findings were based on a single case of Stargardt disease, FAF and NIR-FAF images did not show above pattern. From this point of view, we speculate this discrepancy might be caused by metabolic difference between lipofuscin and melanin in the end-stage Stargardt disease.

In conclusion, FAF was useful in visualizing retinal flecks even though the end-stage Stargardt disease.

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