

A rare and unusual case of unilateral foveal sparing in Stargardt disease

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Abstract: We present a case of Stargardt disease with unilateral good visual acuity in a 68-year-old man. His best corrected visual acuity was 0.9 in the right eye and 0.5 in the left eye. Fundus examination revealed atrophic macular area surrounded by numerous ill-defined grayish flecks in both eyes. Moreover, normal fundus coloration was maintained at the fovea in the right eye. Optical coherence tomography demonstrated irregular thickening of the retinal pigment epithelium, and the photoreceptor inner segment/outer segment (IS/OS) line was disrupted. However, IS/OS line was relatively preserved at the fovea in the right eye. Our findings may contribute to a better understanding of this rare condition.

Keywords: Stargardt disease, foveal sparing, visual acuity, optical coherence tomography, photoreceptor inner segment/outer segment junction.

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, the best corrected visual acuity (BCVA) rapidly deteriorates and usually decreases, by the second decade of life, to below 0.1 or “counting fingers” as a measure of visual acuity [2]. Although the BCVA prognosis is poor, there have been published reports of some patients having good prolonged BCVA despite late-stage disease [2–4]. In addition, several reports have described those conditions, aided by examination with optical coherence tomography (OCT) [2–4]. Here in, we describe a case of Stargardt disease with unilateral good BCVA.

CASE REPORT

A 68-year-old Japanese man was referred to our clinic for a 4-year history of bilateral unclear vision. His medical history was unremarkable, and there was no family history of ocular disease. On ophthalmic examination, his BCVA was 0.9 in the right eye and 0.5 in the left eye, both anterior segments were normal, and the ocular pressures were normal. Ophthalmoscopy of both eyes revealed atrophic macular area surrounded by numerous ill-defined grayish flecks (Figures 1A and 1B). Moreover, normal fundus coloration was maintained at the fovea in the right eye (Figure 1A arrows), while diffuse chorioretinal atrophy with foveal involvement was observed in the left eye (Figure 1B).

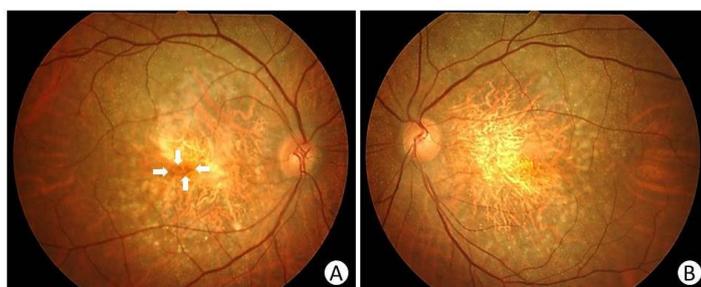


Fig.1 Fundus photographs of the (A) right and (B) left eyes
Marked chorioretinal atrophy surrounded by grayish flecks is visible.
Note the fovea is spared in the right eye (arrows).

Fluoresce in angiography revealed hyperfluorescence corresponding to chorioretinal atrophy in both eyes

(Figures 2A and 2B). However, normal-appearing fovea was detected in the right eye (Figure 2A arrows).

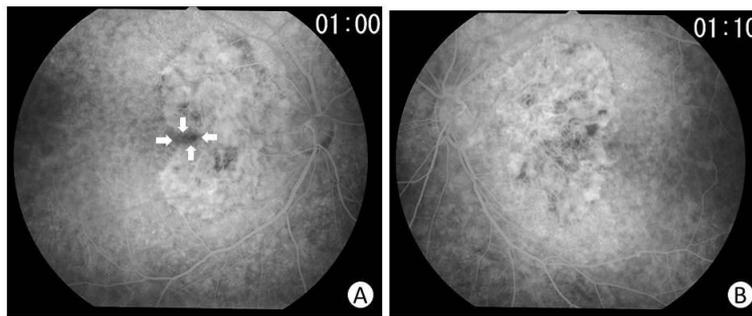


Fig.2: Fluoresce in angiography of the (A) right and (B) left eyes
Note hyperfluorescence in both eyes, while normal-appearing fovea was detected in the right eye (arrows).

OCT (CIRRUS; Carl Zeiss, Germany) demonstrated irregular thickening of the retinal pigment epithelium (Figures 3A and 3B), and the photoreceptor inner segment/outer segment (IS/OS) line was

disrupted. However, IS/OS line was relatively preserved at the fovea in the right eye (Figures 3A and 3C, arrows). High magnification of OCT images confirmed the above findings (Figures 4A and 4B).

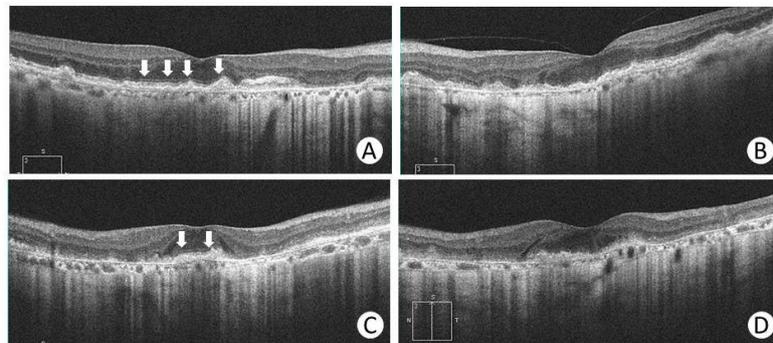


Fig.3: Optical coherence tomography (OCT) images of the right (A, C) and left (B, D) eyes in horizontal (A, B) and vertical (C, D) directions

Note marked attenuation of the IS/OS line in both eyes, while IS/OS line was relatively preserved at the fovea in the right eye (arrows).

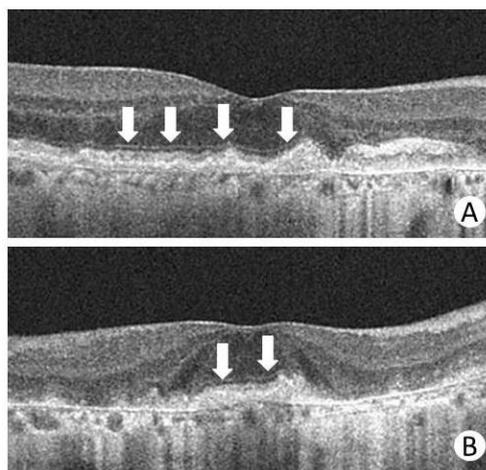


Fig.4: High magnification of OCT images of the right in horizontal (A) and vertical (B) directions
Note the preserved IS/OS line at the fovea (arrows).

Based on these collective findings, we diagnosed our patient with a unilateral foveal-sparing form of Stargardt disease.

DISCUSSION

Patients with Stargardt disease usually have a poor visual prognosis. Rotenstreich et al. [5] evaluated BCVA in 365 patients with Stargardt disease. According to their report, 82 of the 365 patients (23%) had 0.5 or better acuity in at least one eye, 64 (18%) had acuity of 0.4 to 0.2, and 199 (55%) had acuity of 0.1 to 0.05, whereas 16 (4%) had worse than 0.05 acuity in each eye. In the patients with visual acuity of 0.5 or better, 59 (72%) had foveal sparing visible on ophthalmoscopic examination.

Using OCT, Ergun et al. [6] described the correlation between photoreceptor morphology and visual acuity. All Stargardt patients with central atrophy had a complete loss of the central photoreceptor layer in the foveal region. Patients without clinically evident central atrophy had an intact photoreceptor layer centrally but had small, focal parafoveal defects. A correlation was detected between visual acuity and transverse photoreceptor loss. Thus, OCT allows quantitative assessment of the photoreceptor layer in patients with Stargardt disease and correlates it with visual acuity.

Some studies have suggested a slower progression of visual loss in older patients with Stargardt disease [5, 7]. In addition, Westeneng-van Haften et al. [7] described the genotype and phenotype of 21 patients with a late-onset (≥ 45 years) Stargardt disease. According to their report, visual acuity was ≥ 0.5 in 24 eyes of 14 patients (59%) owing to foveal sparing. They found a single heterozygous *ABCA4* variant in 11 patients (52%), 2 compound heterozygous variants in 8 patients (38%), and a homozygous variant in 2 patients (10%). Thus, late-onset Stargardt disease is at the mild end of the spectrum of retinal dystrophies caused by *ABCA4* mutations.

Recently, OCT imaging has been providing insights into the retinal architectural changes that occur in Stargardt disease with preserved foveal structure [2–4]. Using OCT, the preserved photoreceptor in the fovea was clearly detected in recent studies, the findings of which were very similar to the results in the present case. Furthermore, Fujinami et al. [3] described clinical and molecular characteristics of patients with the foveal-sparing phenotype of Stargardt disease. According to their report, there was a higher prevalence of the variant mutation (p.Arg2030Gln) with foveal sparing compared to the group with foveal atrophy. Interestingly, they also found that this phenotype was

associated with a later onset of disease and better visual acuity.

Our findings were based on a single case; additional studies including long-term follow-up, additional cases, and genetic examination are necessary. Finally, OCT was useful in visualizing preserved IS/OS line in a patient with foveal-sparing form of Stargardt disease.

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