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Case Report

# Fundus autofluorescence imaging and optical coherence tomography analysisin a patient with adult-onset foveomacularvitelliform dystrophy

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**Abstract:** We present a case of adult-onset foveomacularvitelliform dystrophy (AFMVD) in a 71-year-old woman. Fundus examination revealed geographic atrophy within the macula. Fundus autofluorescence (FAF) imaging showed clearly defined hypofluorescent lesions corresponding to the atrophic lesion. Near-infrared FAF (NIR-FAF) imaging showed central hyperfluorescent lesions corresponding to the hypofluorescent dark area examined by FAF. Optical coherence tomography (OCT) revealed overall thinning of the neurosensory retina with disruption of the photoreceptor inner segment/outer segment interface. Retinal changes were more easily defined on OCT and FAF images than on color photographs. OCT and FAF images were useful in a patient with AFMVD.
**Keywords:** Adult-onset foveomacularvitelliform dystrophy, fundus autofluorescence imaging, optical coherence tomography

## INTRODUCTION

Adult-onset foveomacularvitelliform dystrophy (AFMVD) is a relatively uncommon macular disease that shares phenotypic features with Best vitelliform macular dystrophy (BVMD)[1]. BVMD is generally diagnosed based on the observation of subretinal deposition of yellowish material within the macula during fundus examination [1]. In BVMD, five stages have been described, based on fundus examination findings: the previtelliform stage (normal macula or subtle alteration of the retinal pigment epithelium (RPE)), the vitelliform stage (a well-circumscribed lesion resembling an egg yolk,0.5 to 2 disc diameters in size), the pseudohypopyon stage(yellow material accumulated inferiorly), the vitelliruptive stage (partial resorption of the material, scrambled-egg lesion), and the atrophic/fibrotic stage (final macular atrophy or fibrosis)[1-4].Several recent reports have described the use of optical coherence tomography (OCT)[1-9] and fundus autofluorescence (FAF) to examine cases of AFMVD or BVMD[1,2,4-9].However, few reports have focused on the FAF of patients with various stages of BVMD [8,9].We describe FAF and OCT findings in a patient with atrophic stage of AFMVD.

## CASE REPORT

A 71-year-old Japanese woman was referred to our clinic for a 3-year history of blurry vision. She had no significant medical history. Her best corrected visual acuity (BCVA) was 0.5 in the right eye and 0.3 in the left eye. Slit-lamp examination showed cortical opacities in both lenses. Ophthalmoscopy revealed geographic atrophy within the maculain both eyes (Fig-1).

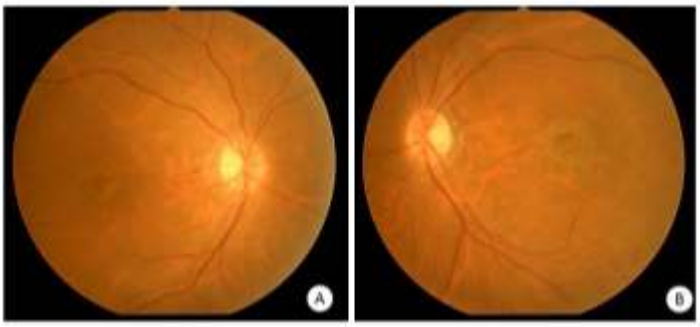


**Fig-1: Fundus photographs of the (A) right and (B) left eyes. Geographic atrophywas observed within the macula.**

FAF (Heidelberg Retina Angiograph 2, Heidelberg Engineering, Heidelberg, Germany) imaging showed clearly defined hypofluorescent lesions corresponding to the atrophic lesion (Fig-2). In addition, some residual dispersed mild hyperautofluorescent material was observed around the atrophic area.

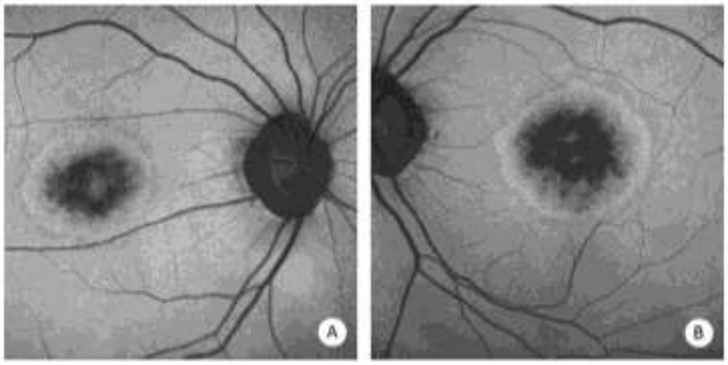

**Fig-2: Fundus autofluorescent (FAF) imaging of the (a) right and (b) left eyes. FAF imaging shows clearly defined hypofluorescent dark area.**

Near-infrared FAF (NIR-FAF) imaging showed central hyperfluorescent lesions corresponding to the hypofluorescent dark area examined by conventional FAF (Fig-3).

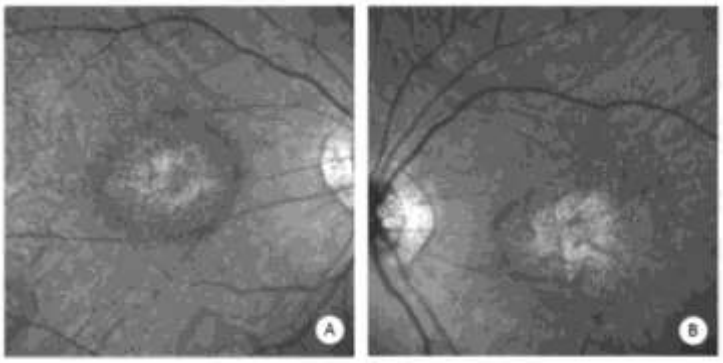

**Fig-3: Near-infrared fundus autofluorescent (NIR-FAF) imaging of the (a) right and (b) left eyes. NIR-FAF imaging shows centralhyperfluorescentarea.**

OCT (DRI OCT-1 Atlantis; TOPCON, Japan) revealed overall thinning of the neurosensory retina with disruption of the photoreceptor inner segment/outer segment (IS/OS) interface (Fig-4, arrowheads).

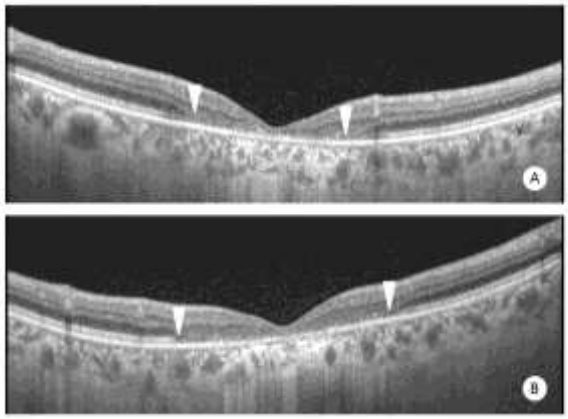

**Fig.-4: Optical coherence tomography (OCT) images of the (A) right and (B) left eyes in the horizontal direction. OCT shows outer retinal atrophy with disruption of theIS/OS interface (arrowheads).**

Based on these collective findings, we diagnosed our patient with atrophic stage of AFMVD.The visual findings did not change during the 10-month follow-up period.

## DISCUSSION

Recently, several reports have described cases of AFMVD or BVMD examined using OCT[1-9].Querques et al. [2] describedAFMVD and suggested that it should be considered as a dynamic process involving alternating phases of material accumulation and reabsorption as it progresses. Querques et al. [3] also described the correlation between BCVA, IS/OS integrity, and stage of the disease. According to their report, BCVA loss has a strong, statistically significant correlation with the presence of focal disruption or diffuse loss of the IS/OS interface, as well as with a more advanced stage of the disease. In our patient with advanced stage, the disruption of IS/OS interface were accompanied by BCVA reduction. Thus, in AFMVD or BVMD, progression of the lesion stage is generally accompanied by IS/OS interface disruption/loss and visual impairment.

Conventional FAF can visualize lipofuscin in the RPE, provides a reliable, noninvasive tool for monitoring the progressive changes of BVMD [8]. In contrast, NIR-FAF appears to correspond to melanin (present in the RPE as well as the choroid) rather than lipofuscin [8]. As such, it provides a different type of information, which may be complementary to FAF. Previous investigations have shown variable patterns of FAF in BVMD, varying from an increased signal, especially visible in the early stages, to a decreased response toward the later stages[8].In the largest case series of BVMD, Parodi et al. [8]analyzed the conventional FAF and NIR-FAF findings of 40 patients. According to their report, six FAF patterns for both conventional FAF and NIR-FAF were identified, including normal, hyperfluorescent, hypofluorescent, patchy, multifocal, and spoke-like patterns. In particular, the examination of the eyes in atrophic stage revealed that five and one eyes revealed a hypofluorescent and a patchypattern, respectively, on conventional FAF, where as NIR-FAF identified three patchy and three hypoautofluorescent patterns. They described the two FAF techniques showed a pattern concordance in four eyes (66%). Furthermore, Parodi et al. [9] described NIR-FAF showed a hypofluorescent pattern in the majority of eyes with subclinical BVMD. This FAF response may reflect either a reduced amount of melanin or an irregular melanin distribution within RPE cells in the subclinical stage.

Although our findings were based on a single case, FAF, NIR-FAF and OCT images were useful in a patient with atrophic stage of AFMVD.

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