Saudi Journal of Ophthalmology (2014) 28, 104–110

Retinal and Choroidal Imaging Update

Multimodal imaging of adult-onset foveomacular vitelliform dystrophy

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Abstract

Adult-onset foveomacular vitelliform dystrophy (AOFVD) is a clinically heterogeneous maculopathy that may mimic other conditions and be difficult to diagnose. It is characterized by late onset, slow progression and high variability in morphologic and functional alterations. Diagnostic evaluation should include careful ophthalmoscopy and imaging studies. The typical ophthalmoscopic findings are bilateral, asymmetric, foveal or perifoveal, yellow, solitary, round to oval elevated subretinal lesions, often with central pigmentation. The lesions characteristically demonstrate increased autofluorescence and hypofluorescent lesions surrounded by irregular annular hyperfluorescence on fluorescein angiography. Optical coherence tomography studies demonstrate homogenous or heterogeneous hyperreflective material between the retinal pigment epithelium and the neurosensory retina. The visual prognosis is generally favorable, but visual loss can occur from chorioretinal atrophy and choroidal neovascularization.

Keywords: Adult-onset foveomacular vitelliform dystrophy, Optical coherence tomography, Imaging, Macula, Pattern dystrophy

http://dx.doi.org/10.1016/j.sjopt.2014.02.001

Introduction

Adult-onset foveomacular vitelliform dystrophy (AOFVD) is a relatively uncommon macular disease first described by Gass in 1974. [1] It generally presents in the fourth to sixth decades of life in patients who are visually asymptomatic or have mild blurring of vision, small central or paracentral scotomas, or mild metamorphopsia.[2] It is often discovered incidentally on routine examinations. Generally, the visual loss is slowly progressive, but almost all patients retain reading vision in at least one eye throughout their lives.

AOFVD is a clinically heterogeneous disease displaying variability in the size, shape, pigmentary changes, and distribution of macular lesions.[3,4] The typical patient has bilateral, asymmetric, foveal or perifoveal, yellow, solitary, round to oval elevated subretinal lesions, often with central pigmentation. The various descriptions in the literature do not make for an easy diagnostic approach, and this disease is often confused with other pigment epithelial alterations.[5,6] Most often, AOFVD is misdiagnosed as age-related macular degeneration, but it can also mimic Best disease. When these patients are referred to a retina specialist for evaluation, few of them are suspected of having AOFVD.

It is therefore important to carefully define the ophthalmoscopic and imaging characteristics of this maculopathy to improve the diagnostic accuracy. This review will discuss different imaging modalities and associated findings that are characteristic of AOFVD.

Clinical examination findings

AOFVD is generally characterized by mild visual loss. In one study of 21 eyes, best-corrected visual acuity ranged from 20/25 to 20/400, with a mean visual acuity of 20/50. 6

Available online 19 February 2014

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Peer review under responsibility of Saudi Ophthalmological Society, King Saud University

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eyes were better than 20/40 and 11 eyes were between 20/40 and 20/63. Only 4 eyes were worse than 20/63.[2] Other studies showed similar distributions.[7]

Renner et al.[8] found that 38 of 75 eyes had normal color testing. The other 37 eyes showed nonspecific errors of variable severity without any typical axis of confusion. Upon visual field testing, 28 of 53 eyes had normal visual fields, whereas 25 demonstrated a central scotoma. An absolute scotoma was observed in only four eyes.

On ophthalmoscopy, AOFVD is characterized by various patterns of unilateral or bilateral, yellow, round to oval elevated subretinal lesions, that often contain a central small, pigmented spot (Fig. 1). Lesions can be barely detectable or up to one disk area in size, are located at the fovea or the perifoveal region, and may be surrounded by small drusen.[2,7] Bilateral lesions were present in 29 of 43 patients in one study[9] and in 11 of 49 patients in another.[7] Another evaluation of 8 patients with AOFVD revealed the presence of a central small, pigmented spot bilaterally in 6 of the 8 patients. In 2 eyes, there were several smaller yellow flecks close to the central lesion and the yellow material was found to fade gradually from the center of the lesion toward its periphery.[10] Importantly, patients may also present later in the course of the disease when the lesions take on different clinical characteristics. Eventually, the lesions may fade, leaving an area of RPE alteration or atrophy.

Complications of the disease that can cause visual loss include choroidal neovascularization in up to one-third of patients, geographic atrophy, and outer retinal atrophy.[11–13] Abrupt visual changes may occur as a result of new onset

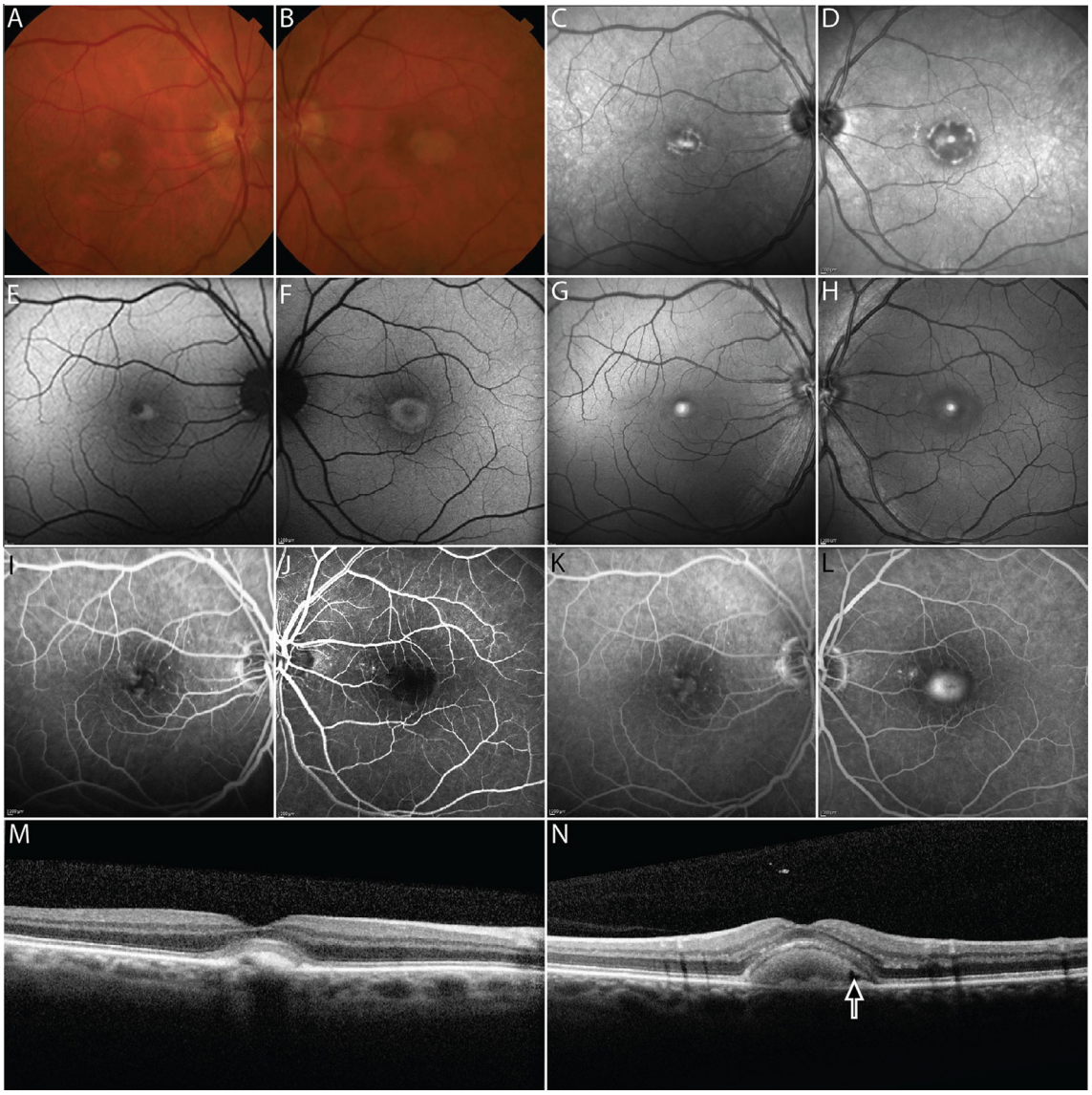

Figure 1. Adult-onset foveomacular vitelliform dystrophy in a 64-year-old man who presented with blurry vision, previously misdiagnosed as age-related macular degeneration. (A and B) Color fundus photographs show bilateral, vitelliform, circular, foveal lesions. (C and D) Infrared imaging shows central white spots surrounded by rings of darker material bordered by mottled outlines of white material. (E and F) The lesions have central hypoautofluorescence surrounded by a ring of hyperfluorescence. (G and H) Red free frames show the central white spots. Fluorescein angiography shows mottled foveal hyperfluorescence with late staining without leakage (I: OD at 01:02, J: OS at 00:30, K: OD at 04:38, L: OS at 4:48). (M) Horizontal spectral-domain optical coherence tomography (SD-OCT) OD shows a disrupted external limiting membrane (ELM) and a mottled and hyperreflective photoreceptor layer that overlies a subretinal hyperreflective dome-shaped lesion. (N) Vertical SD-OCT scan of the left eye (right side of the image is superior macula), showing a mottled ELM and inner segment/outer segment junction, which overlies a heterogeneously hyperreflective subretinal material. The arrow points to an optically empty zone superiorly within the subretinal space, signifying a subtle pseudohypopyon.