

**CASE REPORT**

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# Adult-onset foveomacular dystrophy

## ABSTRACT

### Objective

To describe a case of adult-onset foveomacular vitelliform dystrophy (AOFVD).

### Method

This is a case report.

### Results

A 22-year-old female presented with painless blurring of vision and metamorphopsia 3 days prior to consultation. There were 2 similar episodes in the past that spontaneously resolved after 2 to 4 weeks. Visual acuity (VA) was 20/50 in the right eye (OD) and 20/40 in the left (OS), both best corrected to 20/25. Dilated-fundus examination revealed a discrete area of mixed hypo- and hyperpigmentation 1 disc diameter over the fovea in OD and a solitary round hypopigmented lesion with a hyperpigmented border 3 to 4 disc diameters on the fovea in OS. Fluorescein angiography (FA) revealed an area of hyperfluorescence surrounded by a rim of hypofluorescence in OD and an area of blocked fluorescence with subtle hyperfluorescence superior to the lesion in OS, both of which did not increase in size and intensity toward the late phases. Optical coherence tomography (OCT) revealed neurosensory detachment in both eyes. Electrooculogram (EOG) was normal with Arden ratio of 0.91. VA returned to 20/25 in both eyes, and repeat fundus photography showed no change in the characteristics of the lesions.

### Conclusion

Differential diagnosis of a hypopigmented macular lesion in the young with self-limited blurring of vision should include AOFVD. FA, OCT, and EOG can help distinguish AOFVD from Best's disease or other similar macular conditions.

**Keywords:** *Adult-onset foveomacular vitelliform dystrophy, Best's disease, Vitelliform macular dystrophy, Bestrophin, Peripherin*

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IN 1905, FRIEDRICH Best first described a yellowish lesion in the macula of 8 young blood-related individuals with good and stable vision. These lesions were later labeled vitelliform because they resembled an egg yolk. Since then, Best's disease or Best's familial macular degeneration has also been known as congenital macular dystrophy, congenital vitelliform cysts of the macula, and hereditary vitelliruptive macular degeneration.<sup>1,2</sup>

In 1974, Gass described a vitelliform-like lesion with a central pigmented spot in his adult patient with slow and progressive vision loss.<sup>1</sup> This was named pseudo-Best, pseudo-vitelliform, adult foveal macular dystrophy of Gass, and finally, adult-onset foveomacular vitelliform dystrophy (AOFVD).<sup>2</sup> Since then, Best's disease and AOFVD have been included in the differential diagnosis in a patient with a yellowish macular lesion. A plethora of reports have been made on Best's disease; however, only a few literature has documented AOFVD.<sup>1,2</sup>

### CASE REPORT

A 22-year-old female consulted for intermittent blurring of vision in both eyes. A year earlier, she had sudden blurring of vision, worse in the right eye, accompanied by seeing straight images as wavy lines. There was no eye pain or redness. These symptoms spontaneously resolved after a month without consultation.

One month prior to consultation, the patient experienced blurring of vision similar to previous episodes, which resolved in 2 weeks. Three days before consulting, she experienced the same symptoms, accompanied by eye redness, photophobia, and tearing of the left eye.

The patient did not smoke tobacco or drink alcohol. Medical history and review of systems were unremarkable. She had no previous eye surgeries or use of any eye medications. She reported that her maternal uncle had similar transient blurring of vision and was allegedly monitored and managed by an ophthalmologist for an unrecalled eye disease.

Visual acuity using the Snellen chart was 20/50 in the right eye (OD) and 20/40 in the left eye (OS), best corrected to 20/25 for both eyes (OU). Metamorphopsia was reported bilaterally at the center of OU on Amsler grid testing. Color vision was 9/10 on Ishihara and mild tritan defect on Farnsworth D15 in OU. Both pupils were round, regular, 2 to 3mm, and briskly reactive to light and accommodation with no afferent pupillary defect. The rest of the anterior-segment findings were normal except for mild conjunctival injection and a 2 x 2-mm corneal epithelial defect with fluorescein uptake in OS. Extraocular motility and visual-field screening were normal.

Funduscopy revealed a discrete round area of mixed hypo- and hyperpigmentation 1 disc diameter in size over the fovea of OD and a solitary hyperpigmented yellow

spot 3 to 4 disc diameters in size obscuring the fovea in OS (Figure 1).

The patient was initially treated with moxifloxacin 4 times a day for 1 week for the corneal abrasion in OS while the macular lesions were placed under observation.

One week after consultation, patient spontaneously regained vision. Uncorrected visual acuity (UCVA) was 20/25 in OU. A superficial corneal scar was present on the inferior quadrant of OS.

Fluorescein angiography (FA) revealed an area of hyperfluorescence surrounded by a patch of hypofluorescence in OD and blocked fluorescence at the fovea corresponding to the subretinal lesion in OS (Figure 2). Vitelliform macular dystrophy was considered in OU.

Optical coherence tomography (OCT) in OD revealed a shallow foveal depression with detachment of the neurosensory retina (Figure 3A). Retinal pigment epithelium (RPE) was intact and thickened at the area of detachment (Figure 3B). The choroid was unremarkable. Central foveal thickness ( $262 \pm 3$  microns) was increased and retinal-map analysis showed thickened fovea (Figure 4).

OCT in OS showed formed but shallow and distorted foveal depression (Figure 5A) with a small area of neurosensory detachment subfoveally on vertical scan (Figure 5B), adjacent to which was a confluent area of high reflectivity involving the RPE. Weak signals from the choroid were noted beneath this area. Central foveal thickness could not be measured accurately and retinal-map analysis was possibly inaccurate (Figure 6).

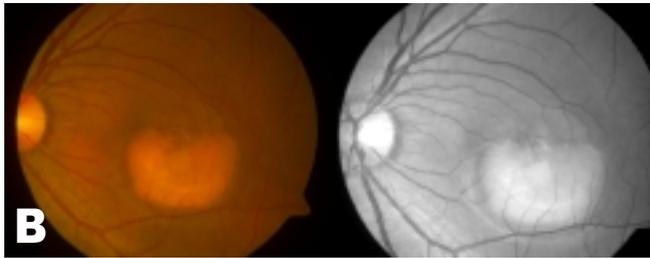
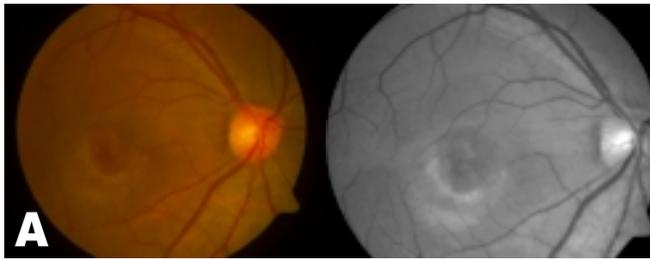
Electrooculogram (EOG) was normal. The Arden ratio was 0.91 with an Arden index of 214% (LP 2515u and DT 1174u) in the right and 234% (LP 1010u and DT 432u) in the left eye (Figure 7).

The patient was diagnosed to have AOFVD and no treatment was given. Three months after the initial consultation, patient reported improvement of metamorphopsia on Amsler with UCVA of 20/25 in OU. Repeat fundus photography showed no change in the characteristics of the lesions.

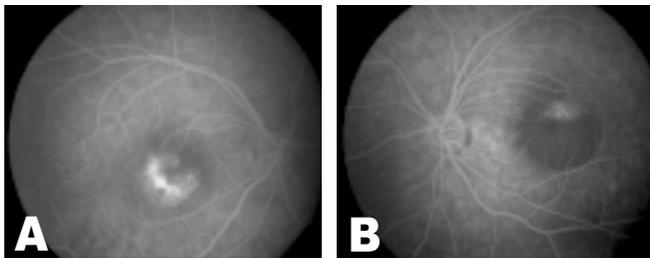
### DISCUSSION

Best's disease presents early in the first 2 decades of life while AOFVD presents later in adulthood in the third to fifth decades.<sup>1</sup> Best's disease is usually asymptomatic or with very minimal blurring of central vision with or without mild metamorphopsia, whereas AOFVD presents with gradual worsening of vision.<sup>1</sup> Our patient was a 22-year-old female previously asymptomatic with transient blurring of vision and mild metamorphopsia.

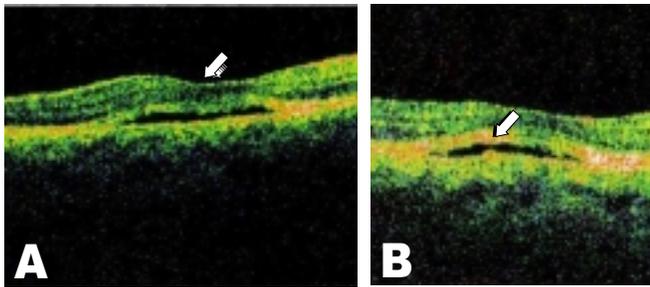
Both Best's disease and AOFVD are autosomal-dominant, with variable expression and incomplete penetrance.<sup>1</sup> Best's disease is caused by mutations in the VMD2 gene, which encodes the protein bestrophin. Bestrophin



**Figure 1.** Fundus photos showed a discrete round area of mixed hypo- and hyperpigmentation 1 disc diameter in size over the fovea of the right eye (A) and a solitary hyperpigmented yellow spot 3 to 4 disc diameters in size obscuring the fovea of the left (B).

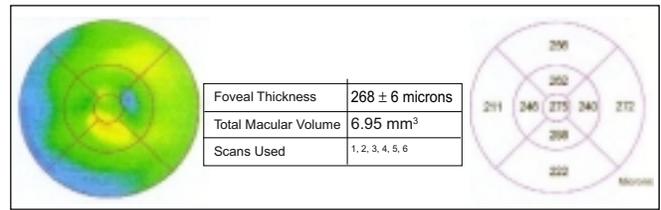


**Figure 2.** Fluorescein angiography showed an area of hyperfluorescence surrounded by a patch of hypofluorescence in the right (A) and blocked fluorescence at the fovea corresponding to the subretinal lesion in the left (B).

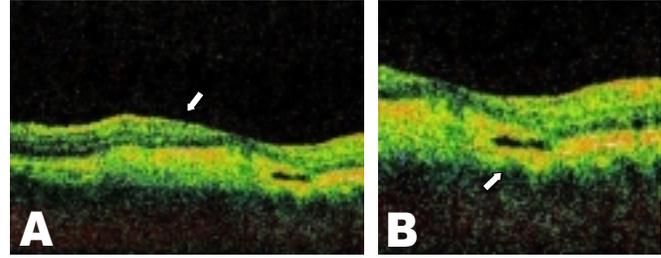


**Figure 3.** Optical coherence tomography of the right eye showed shallow foveal depression (→) with a detachment (solid black space) of the neurosensory retina (A). Retinal pigment epithelium (⇨) was intact and noted to be thickened at the area of detachment (B).

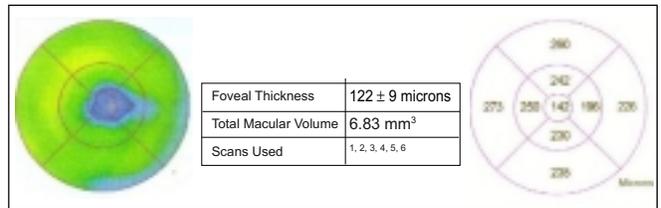
functions as a transmembrane chloride channel and its absence results to abnormal lipofuscin accumulation in the fovea.<sup>2</sup> On the other hand, the specific gene for AOFVD remains unknown. Few cases of AOFVD share a common mutation in the VMD2 gene with Best's disease.<sup>3</sup> Another possible cause has been suggested in the human retinal-degeneration-slow (RDS) gene that encodes peripherin.



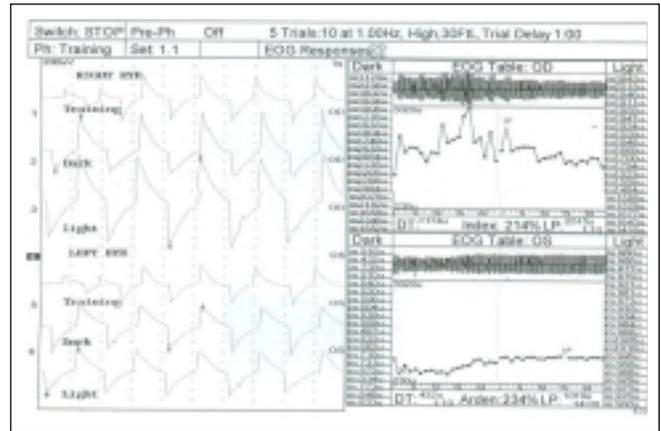
**Figure 4.** Retinal-map analysis of the right eye showing thickened fovea.



**Figure 5.** Optical coherence tomography of the left eye showed formed but shallow and distorted foveal depression (A). A small area of neurosensory detachment was observed subfoveally on vertical scan, adjacent to which was a confluent area of high reflectivity involving the RPE (B).



**Figure 6.** Retinal-map analysis of the left eye.



**Figure 7.** Normal electrooculogram consistent with adult-onset foveomacular dystrophy.

Peripherin is a glycoprotein in the outer segments of photoreceptors responsible for maintaining the structural integrity of rods and cones. Eighteen percent of patients with AOFVD have mutations in the peripherin/RDS gene.<sup>3</sup>

Both diseases are heterogenous and pleomorphic. Best's disease usually presents as a solitary lesion while AOFVD may be multifocal.<sup>2</sup> In 1971, Deutman classified

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the evolution of Best's disease in different stages, which was later modified by Mohler and Fine in 1981. These include previtelliform, vitelliform, pseudohypopyon, vitelliruptive, and atrophic or scarring.<sup>1-2</sup>

AOFVD is similar to Best's as a slightly elevated yellow subfoveal lesion, but is smaller, usually 1/3 disc diameter, more symmetrically round or oval with a central pigmented spot.<sup>2</sup> AOFVD does not necessarily progress through the 5 stages of Best's and if it does, the end stage is usually RPE atrophy; it does not progress to hypertrophic scars or choroidal neovascularization.<sup>1</sup> Our patient presented with pseudohypopyon configuration in OS and an atrophic configuration in OD.

FA shows similar pattern for both AOFVD and Best's. In the vitelliform stage, there is central hypofluorescence at the macula due to the blockage of choroidal fluorescence by the yellowish subretinal fluid with slight hyperfluorescence of the RPE surrounding the lesion forming an irregular ring of hyperfluorescence.<sup>1-2</sup> Our patient had a ring of slight hyperfluorescence in OD and blocked fluorescence in OS, consistent with the pseudohypopyon and atrophic foveal RPE of the right and left eyes.

OCT of Best's disease reveals splitting of the RPE choriocapillaris complex under the fovea—defining an optically clear space of central serous detachment of the RPE.<sup>5</sup> This undergoes variations in the different stages of

Best's disease. In AOFVD, there is a highly reflective subretinal fusiform thickening of the RPE due to the vitelliform lesion between the RPE and photoreceptor layer.<sup>3</sup> Neurosensory detachment of both eyes was considered, consistent with Best's disease.

In Best's disease, the ERG is normal but the EOG is severely abnormal, even in normal carriers of the disease, due to the blockage effect by the vitelliform material.<sup>2</sup> However, in AOFVD, EOG is normal to slightly subnormal<sup>1</sup> as demonstrated in this patient.

In summary, AOFVD is similar to Best's disease but has its distinctive features and natural history. Several diagnostic tests can be performed to help differentiate the 2 conditions.

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