

Evolution of Spectral Domain Optical Coherence Tomography changes in Adult Onset Vitelliform Dystrophy

Muna Bhende, Deepak Agarwal, KP Mohana

Shri Bhagwan Mahavir Vitreoretinal Services, Sankara Nethralaya, Chennai, Tamil Nadu, India

Correspondence: Muna Bhende, Senior Consultant, Department of Retina and Vitreous, Shri Bhagwan Mahavir Vitreoretinal Services, Medical Research Foundation, Chennai, Tamil Nadu, India, e-mail: drmuna@snmail.org

INTRODUCTION

Vitelliform macular dystrophy is an autosomal dominant disorder with variable penetrance and expressivity. It was first described by Friedrich Best¹ in 1905 with a complete description of various stages of this disease. Adult-onset vitelliform dystrophy, which was first described by Gass² in 1974, differs from classic vitelliform dystrophy in that the vitelliform lesion may have a variable evolution. It manifests with bilateral mild to moderate visual loss of late onset. Electrooculogram (EOG) is normal to slightly abnormal. It is an important differential diagnosis for wet AMD where antiVEGF therapy has little or no benefit.³⁻⁵

Optical coherence tomography (OCT) is a useful tool to evaluate and document different stages of this disease. In this report, we documented OCT changes in a patient through different stages of the disease over a follow-up of 10 months.

CASE REPORT

A 58-year-old male came to our outpatient department with complaints of painless gradual progressive diminution of vision in both eyes for last 6 months and a referral diagnosis of wet age-related macular degeneration. On examination, his best corrected visual acuity was OD 6/18, N12 and OS 6/24, N18. Anterior segment examination was within normal limits. Fundus examination revealed retinal pigment epithelium (RPE) atrophic changes in the macula in OD and a vitelliform lesion of about 1 disk diameter (DD) in the macula in OS. There was no subretinal hemorrhage or fluid. Fundus fluorescein angiography (FFA) showed transmission defects in the macula in OD and blocked fluorescence in the area of the vitelliform lesion in OS (Fig. 1). Electroretinogram (ERG) was within normal limits with normal scotopic and photopic responses in both eyes (Fig. 2). Multifocal ERG (MFERG) showed reduced central ring responses and normal paracentral and peripheral ring responses with loss of foveal peak in both eyes (Figs 3 and 4). Electrooculogram (EOG) showed subnormal Arden's ratio. OD: 1.80; OS: 1.63 (Fig. 5). A diagnosis of vitelliform dystrophy was made, possibly adult onset, in view of the absence of a documented fundus pathology earlier on.

Optical coherence tomography (3D-OCT 1000; Topcon, Tokyo, Japan) was done which showed a normal foveal dip, hyperreflective thickened RPE with focal areas of RPE atrophy and focal disruption of the inner segment and outer segment (IS/OS) interface in the foveal region in OD (Fig. 6). OS showed a thickened hyperreflective lesion at and below the retinal pigment epithelium elevating the retina in the macular region (Fig. 7). It also showed focal disruption of IS/OS interface in the fovea. All other intraretinal layers were normal in both eyes.

Since there was no evidence of active choroidal neovascular membrane, the patient was asked to come for a follow-up after 6 months and monitor his vision on a regular basis.

The patient was reexamined after 5 months. He maintained the same best corrected visual acuity and anterior segment findings were also unchanged. OD fundus was unchanged, while in OS fundus the vitelliform lesion now showed a picture similar to the vitelliruptive stage. OCT was done which showed changes in OD as described in previous visit along with a decrease in thickening of RPE layer (Fig. 6). OS showed reduction in the height of the lesion compared to the previous visit with thickened RPE and few cystic spaces (Fig. 7). It also showed focal RPE atrophy along with focal disruption of (IS/OS) interface. All

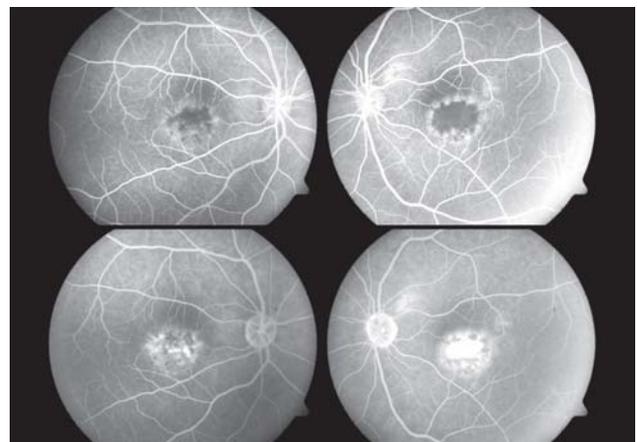
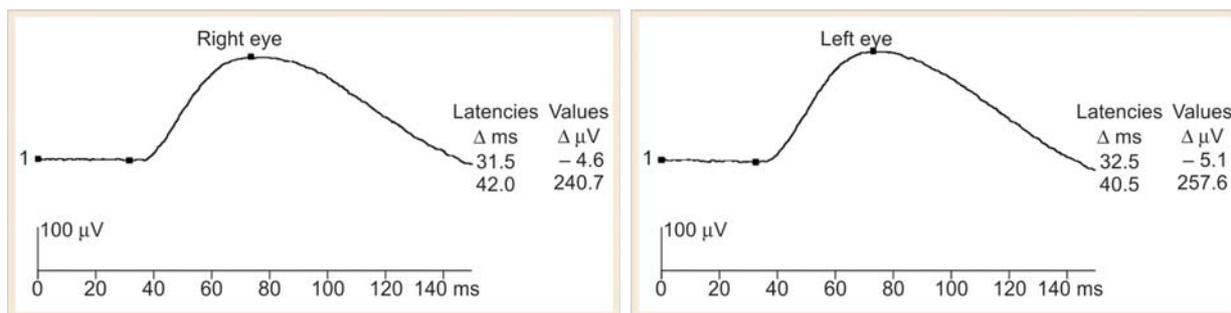


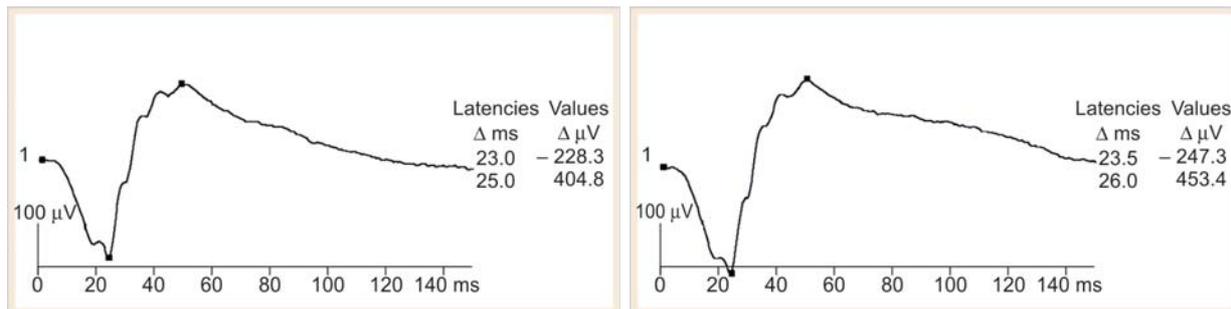
Fig. 1: FFA picture early phase showing transmission defects in right eye and central blocked hypofluorescence surrounded by ring of irregular hyperfluorescence in left eye (top). Late phase showing hyperfluorescence due to staining in both eyes (bottom)

Ganzfeld Summary

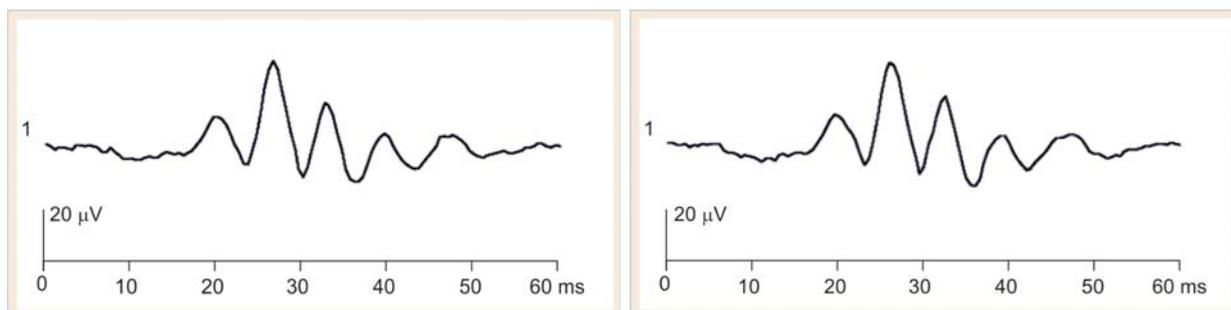
Rod Response



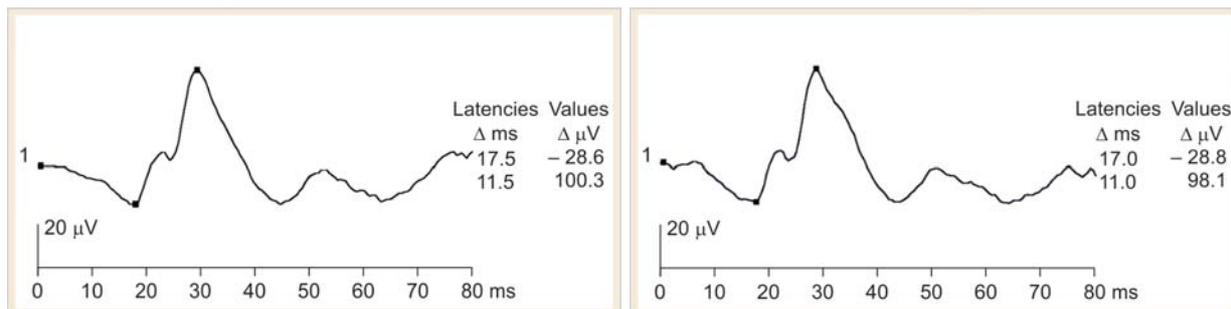
Maximal Response



Oscillatory Potentials



Cone Response



Field view

Field view

30 Hz Flicker

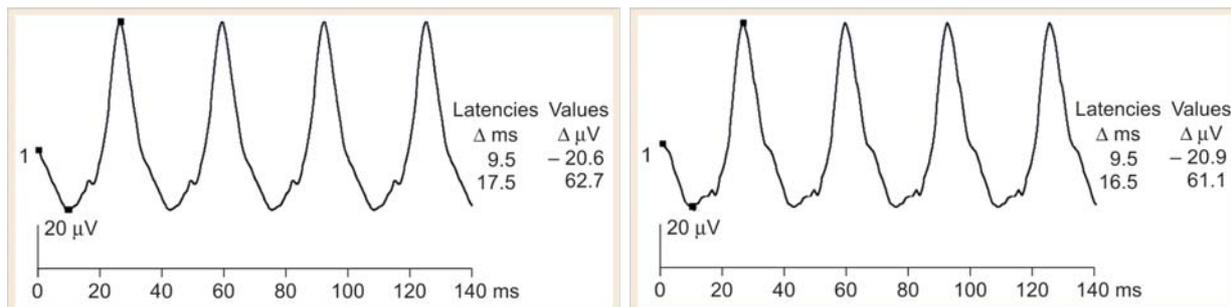


Fig. 2: ERG showing normal scotopic and photopic responses in both eyes (Courtesy: Ganzfeld summary)

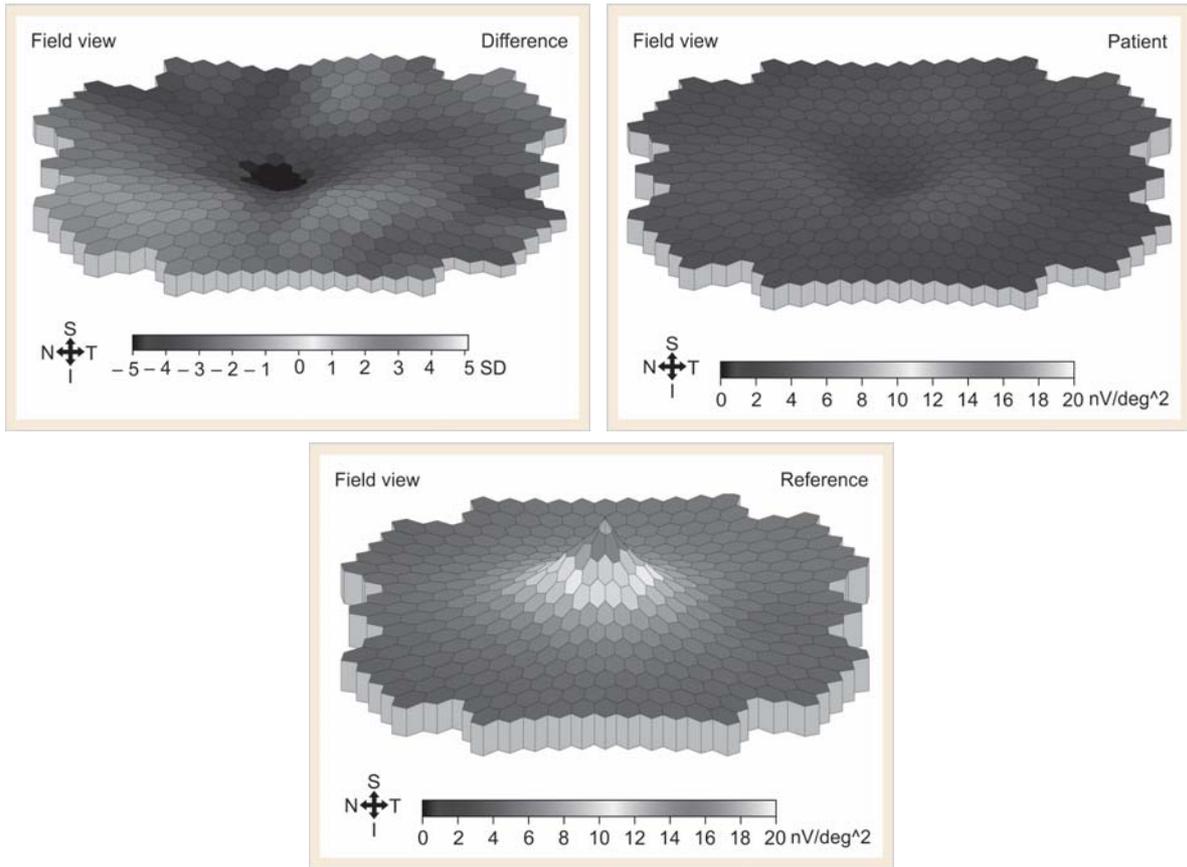


Fig. 3: MFERG showing loss of foveal peak in right eye

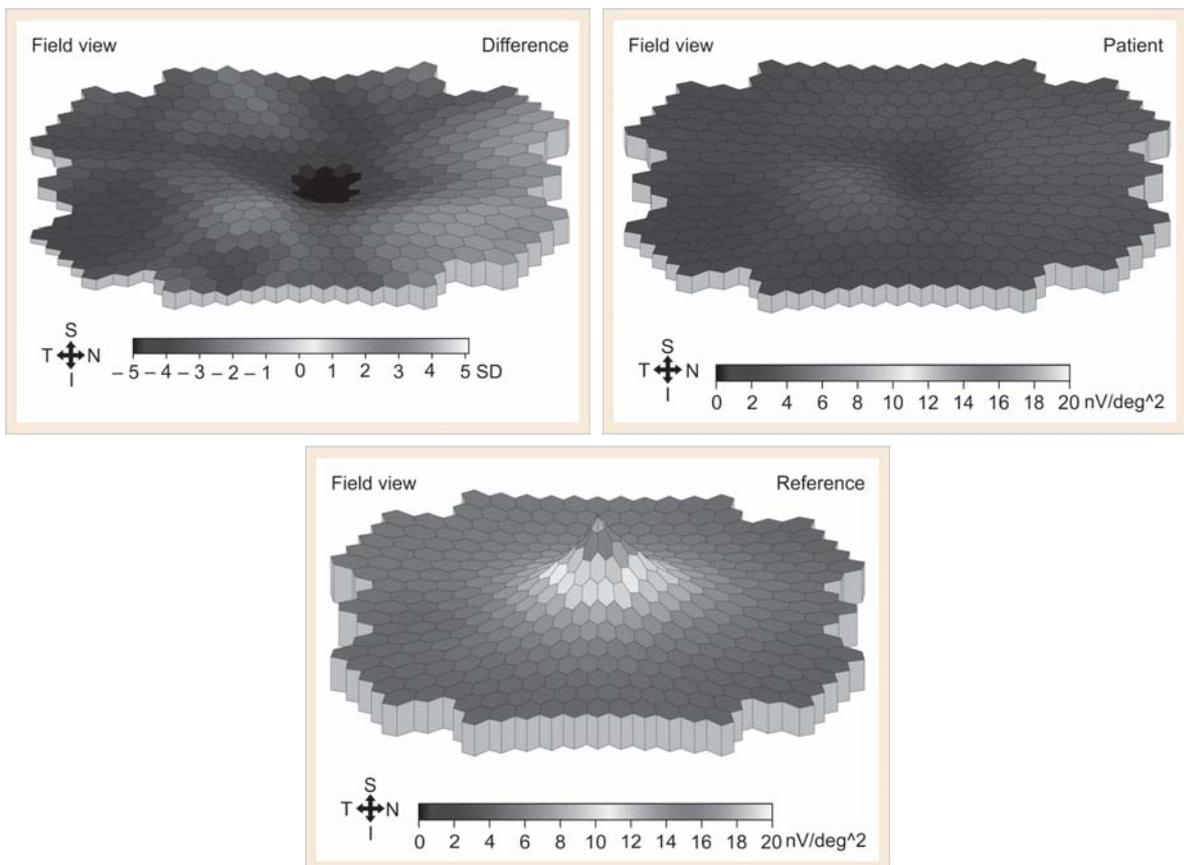


Fig. 4: MFERG showing loss of foveal peak in left eye

the other intraretinal layers were normal. There was no evidence of choroidal neovascular membrane or subretinal fluid. Patient was asked to continue follow-up.

The last visit was 10 months after initial presentation. He maintained the same best corrected visual acuity in OD while OS had a drop from 6/24 on first visit to 6/36. Anterior segment findings were unchanged. On fundus examination clinically the lesion seem to be in the atrophic stage. OCT was done which showed normal foveal contour with focal RPE atrophic changes in both the eyes (Figs 6 and 7). It also showed focal disruption of IS/OS interface in both the eyes. All the other intraretinal layers appeared normal. There was no choroidal neovascular membrane or subretinal fluid. The patient was advised routine follow-up.

DISCUSSION

Vitelliform macular dystrophy has been described to progress through five stages⁶ according to fundus examination findings which are (1) Previtelliform stage, (2) Vitelliform stage, (3) Pseudohypopyon stage, (4) Vitelliruptive stage and (5) Atrophic stage. Adult-onset vitelliform dystrophy may have a variable evolution and may not progress through all these stages.

There have been few previous reports⁷⁻¹⁰ which have described the OCT findings in vitelliform dystrophy but this report is unique in that it presents sequential OCT changes in

the same eye of the same patient through different stages of vitelliform macular dystrophy.

Querques G et al⁸ have described the OCT findings in different stages of vitelliform dystrophy in different patients of their case series. They have described previtelliform lesion as having a thicker layer between the RPE and IS/OS interface. In vitelliform to vitelliruptive stage, the vitelliform material was visualized as a highly reflective lesion located between the hyporeflective outer nuclear layer and the hyperreflective RPE layer, associated or not with an optically empty lesion. Atrophic stage was characterized by hyperreflective mottling on the RPE layer along with thinning of all the retinal layers and diffuse loss of the IS/OS interface.

In a large series of 90 eyes by Freund et al,¹⁰ 12 eyes were noted to have spontaneous regression of vitelliform lesions. Visual acuity decreased along with the regression of the lesion in nine eyes, three showed improvement. All showed varying degrees of atrophic changes on the SD OCT, with thinning and

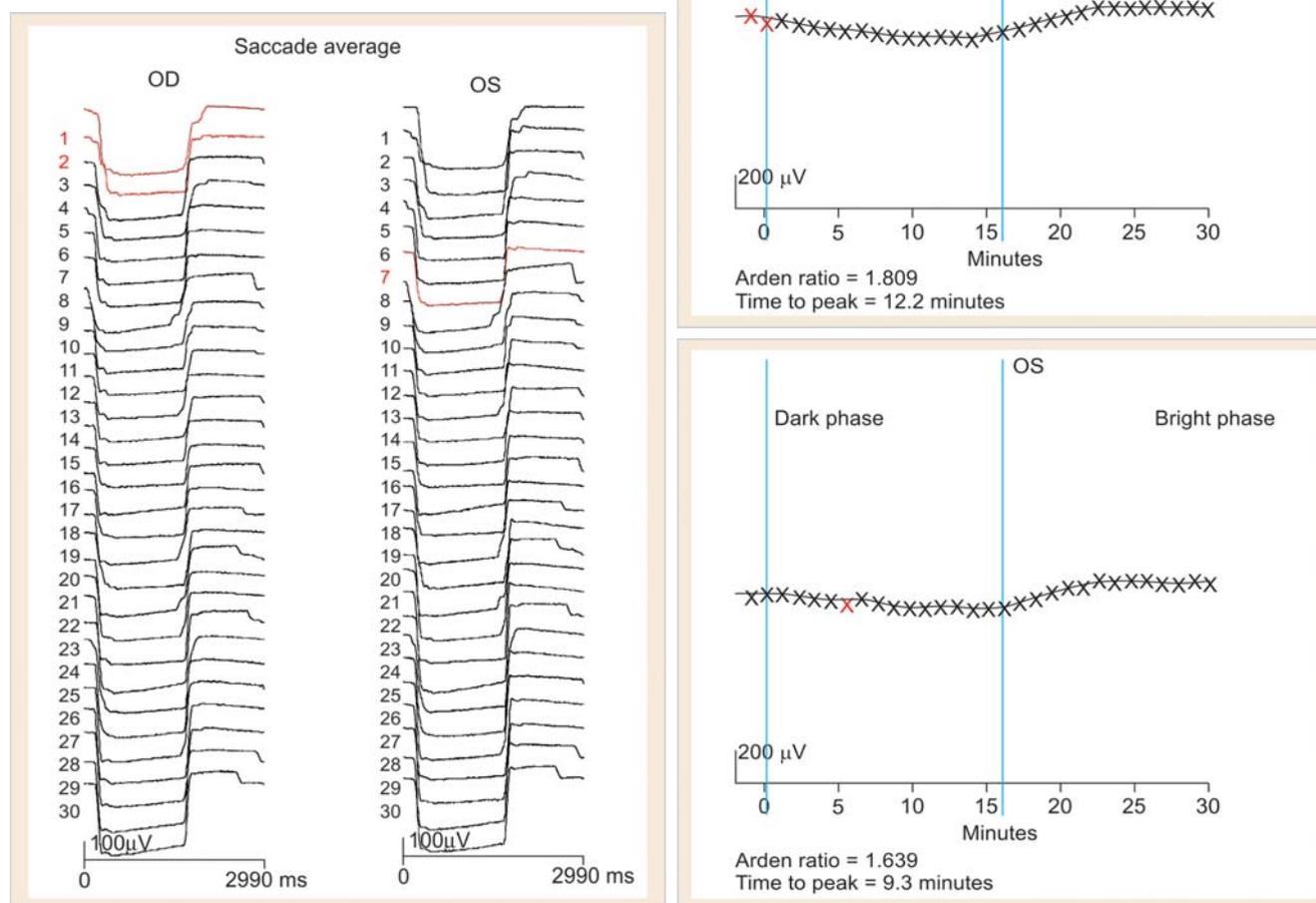


Fig. 5: EOG showing subnormal Arden's ratio in both eyes

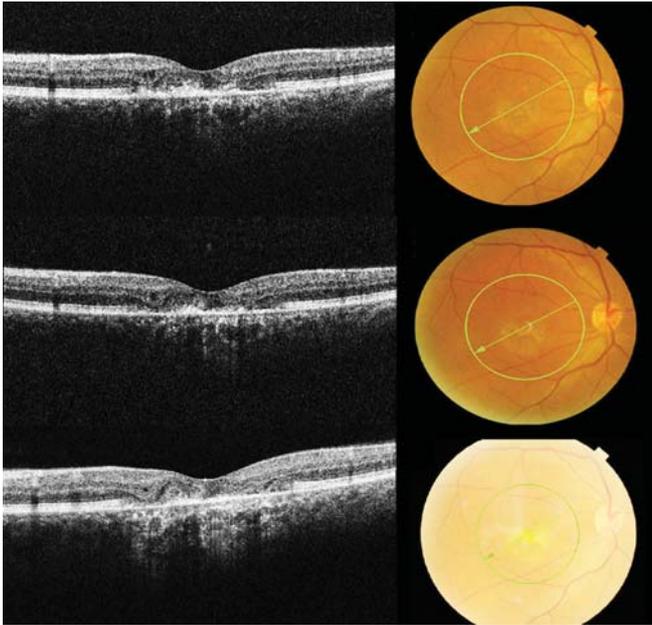


Fig. 6: SD-OCT of right eye showing hyperreflective thickened RPE with focal areas of RPE atrophy and focal disruption of the IS/OS interface in the foveal region (top), decrease in thickening of RPE layer on next visit (middle), focal RPE atrophy along with focal disruption of (IS/OS) interface on last visit (bottom)

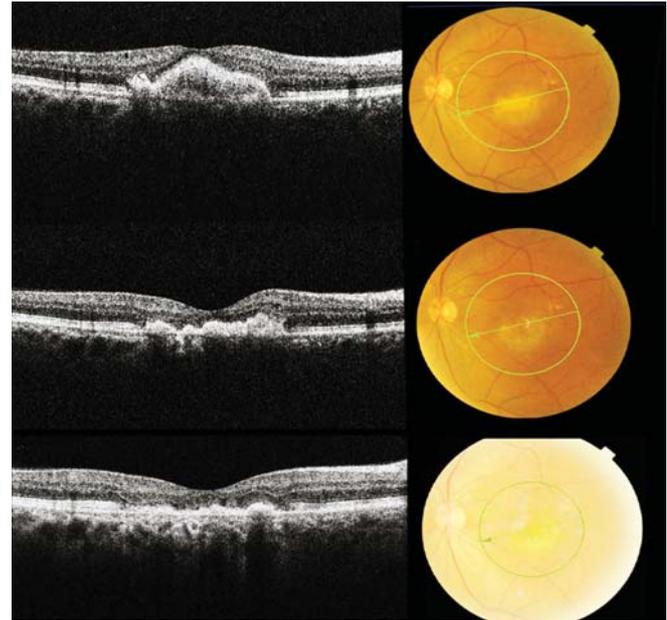


Fig. 7: SD-OCT of left eye showing thickened hyperreflective lesion at and below the retinal pigment epithelium elevating the retina in the macular region with focal disruption of IS/OS interface in the foveal region (top), reduction in the height of the lesion on next visit with thickened RPE and few cystic spaces (middle), normal foveal contour with focal RPE atrophic changes on last visit (bottom)

focal loss of outer nuclear layer, IS-OS junction disruption and thinning or focal loss of the RPE band.

They also noted intraretinal migration of the subretinal material. OCT is a useful tool to monitor and document the intraretinal changes through different stages of this disease. It allows *in vivo* visualization of the intraretinal changes especially the localization of the vitelliform material, retinal pigment epithelial changes and photoreceptors IS/OS junction which is important in correlating the changes due to disease with visual acuity. It is also useful in detecting the development of complications of the disease such as choroidal neovascular membrane.

CONCLUSION

Here, we describe a case of vitelliform macular dystrophy which was seen at three different stages of the disease. It led to development of RPE atrophic changes with focal IS/OS junction disruptions documented on OCT.

REFERENCES

1. Best F. Uber eine hereditaere makulaaffection; Beitrage zur vererbungslehre. *Zschr Augenheilk* 1905;13:199-212.
2. Gass JDM. A clinicopathologic study of a peculiar foveomacular dystrophy. *Trans Am Ophthalmol Soc* 1974;72: 139-56.

3. Montero JA, Ruiz-Moreno JM, De La Vega C. Intravitreal bevacizumab for adult-onset vitelliform dystrophy: A case report. *Eur J Ophthalmol* Nov-Dec, 2007;17(6):983-86.
4. Lee JY, Lim J, Chung H, Kim JG, Yoon YH. Spectral domain optical coherence tomography in a patient with adult-onset vitelliform dystrophy treated with intravitreal bevacizumab. *Ophthalmic Surg Lasers Imaging* May-June 2009;40(3): 319-21.
5. Gallego-Pinazo R, Dolz-Marco R, Pardo-López D, Arevalo JF, Díaz-Llopis M. Primary intravitreal ranibizumab for adult-onset foveomacular vitelliform dystrophy. *Graefes Arch Clin Exp Ophthalmol*, Oct 8, 2010.
6. Gass JDM. Stereoscopic atlas of macular diseases. Diagnosis and treatment. Fourth Edition. The CV Mosby Co St Louis, 1997;316-24.
7. Benhamou N, Souied EH, Zolf R, Coscas F, Coscas G, Soubrane G. Adult-onset foveomacular vitelliform dystrophy: A study by optical coherence tomography. *Am J Ophthalmol* 2003;135:362-67.
8. Querques G, Regenbogen M, Quijano C, Delphin N, Soubrane G, Souied EH. High-definition optical coherence tomography features in vitelliform macular dystrophy. *Am J Ophthalmol* Oct 2008;146(4):501-07.
9. Puche N, Querques G, Benhamou N, Tick S, Mimoun G, Martinelli D, et al. High-resolution spectral domain optical coherence tomography features in adult onset foveomacular vitelliform dystrophy. *Br J Ophthalmol* Sep 2010;94(9):1190-96.
10. Freund KB, Laud K, Lima LH, Spaide RF, Zweifel S, Yannuzzi LA. Acquired Vitelliform Lesions: Correlation of clinical findings and multiple imaging analyses. *Retina*, Jan 2011;31(1):13-25.