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Adult Onset Foveolar Vitteline Dystrophy

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ABSTRACT

Adult onset foveo-macular vitelliform dystrophy is a relatively uncommon condition and often misdiagnosed, as the true vitelliform dystrophy is rare in adults. We describe one case of AOFVD; the case underwent a complete ophthalmic examination, fluorescein angiography, systemic evaluation including physical examination and laboratory examination. We did not find any associated systemic disease in our case in spite of extensive investigations.

INTRODUCTION

Adult onset Foveo-macular vitelliform dystrophy is a condition that presents classically as bilateral, symmetrical, greyish yellow round or oval lesions within the macular area best known as 'poached egg' appearance [1]. These lesions are mildly elevated and are one-third to half disc diameter in size. The onset of disease is usually between 30 and 50 years of age with variable genetic inheritance, although some have suggested an autosomal dominance inheritance pattern. The mutations were observed in the BEST1 and PRPH gene respectively. Patients with AOFVD typically present with symptoms of mild blurred vision or normal vision and mild metamorphopsia and a mild red-green dyschromatopsia seen in later stages of the disease [2].

Results of diagnostic testing show a normal or mildly subnormal electroretinogram (EOG) [3]. Fluorescein angiography (FA) shows a hypo fluorescent area corresponding to vitelliform lesion and surrounding ring of hyper fluorescence [4]. Optical Coherence Tomography (OCT) show the vitelliform lesion as being located in the retinal pigment epithelium (RPE) layer or between the RPE and photoreceptor layer. Vitelliform macular dystrophy is a disease of the retinal pigment epithelium which in later stages known as 'scrambled egg appearance, may lead to chorio retinal atrophy [1].

In 1977 Fisherman et al described 3 patients above the age of 45yrs with bilateral vitelliform lesions with normal EOG findings and visual acuity of 20/100 or better were termed as 'pseudo vitelliform macular degeneration'. On fluorescein angiography found a hyper fluoroscent area around the fovea and was hypothesized due to leakage from perifoveolar capillaries. The authors thus emphasised on the use of EOG to differentiate between pseudovitelliform dystrophy from Best's disease [5].

Kingham and Lochen described 6 cases of vitelliform dystrophy with normal EOGs. Angiography in their cases revealed hyper fluorescent areas due to pigment epithelial defect thus making the emphasis on leakage from choroid rather than perifoveal capillary network [6].

Gass studied 9 cases of what he termed 'peculiar foveolar macular dystrophy', having bilateral, symmetrical, raised yellow sub retinal lesions of 1/3rd disc diameter size with central pigment spot. The onset was primarily in 30 to 50 years of age group, with symptoms of progressive slight blurring of vision and metamorphopsia. EOG readings were found to be sub-normal with fluorescein angiography showing hypo fluorescent lesion or a hyper fluorescent ring with central foveolar hypo fluorescence. The pathology was found in the retinal pigment epithelium, with some relationship of this entity to family drusen [7].

CASE

A 60 year old female patient presented on April 2016 with complaints of diminution of vision both eyes for 5-6 years and was diagnosed as a case of central confluent drusen with presbyopia at a private hospital and was prescribed with refractive correction but the condition worsen. Examination of both the eyes showed a Best-corrected visual acquity of 6/12 in Right eye and 6/9 in the left eye with normal adnexa, cornea and conjunctiva, anterior chamber and IOP. Fundoscopy revealed a central confluent drusen with pigmentary dystrophy at the macular area. The Red-free

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image showed a mottled hypo pigmented spots in the macular region with 3-4 hyper pigmented spots on the inferior aspect of the lesion.

Fluorescein angiography showed a hyper fluorescent lesion in the macular region approximately the size of one-disc diameter with 3-4 hypo fluorescent spots present inferiorly. The OCT showed hypo reflective lesion beneath the RPE in both the eyes at the macular region.

Routine blood investigations showed deranged lipid profile of increased LDL 0f 114 mg/dl and Kidney function test with increased uric acid (6.7mg/dl) and alkaline phosphatase (181 U/l). Electro-oculogram was in normal limits.

She was diagnosed as a case of adult onset foveo-macular vitelliform dystrophy and was advised to undergo 6-monthly follow-up for any progression **(Figures 1-8)**.

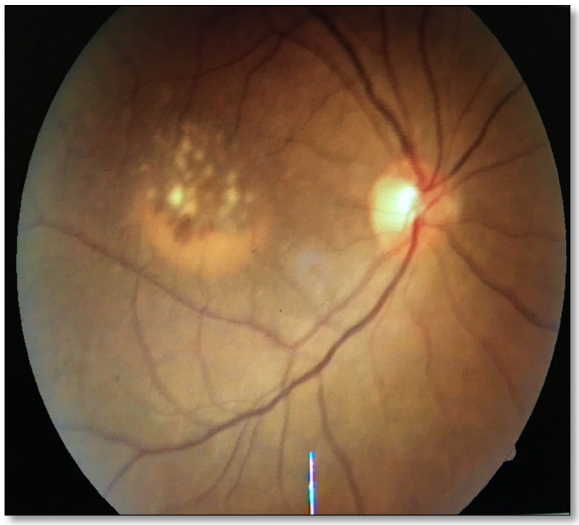

Figure 1. Fundus right eye with mottled hypopigmented leisions on macular region.

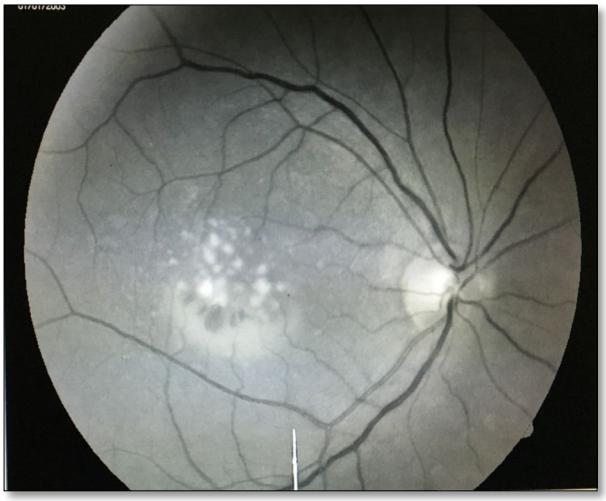

Figure 2. Red free photo of right eye showing hypo pigmented lesion on the macular region.

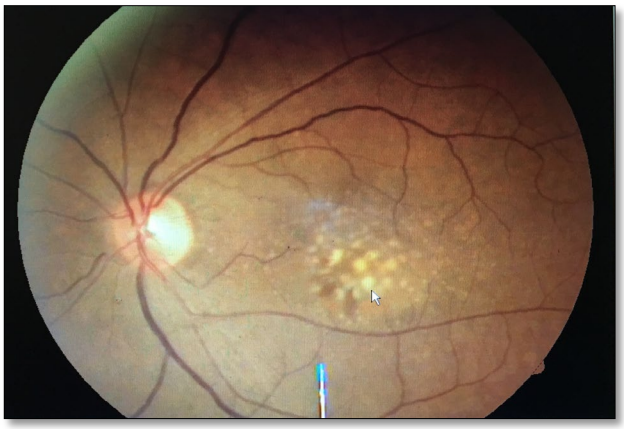

Figure 3. Fundus of left eye showing mottled hypo pigmented lesion on macular region.

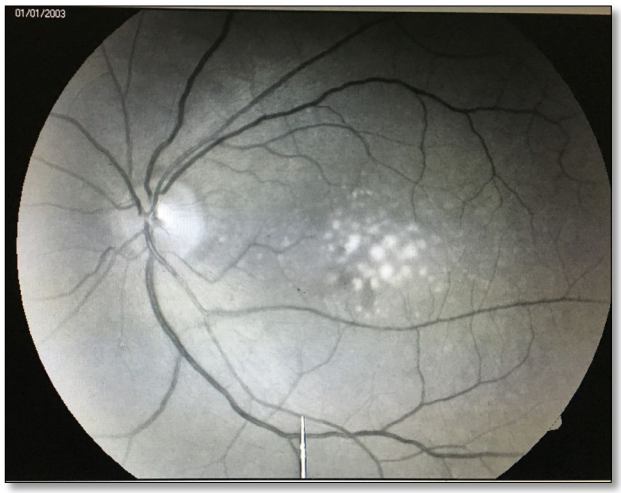

Figure 4. Red free image showing hypopigmented lesion in the macular region.

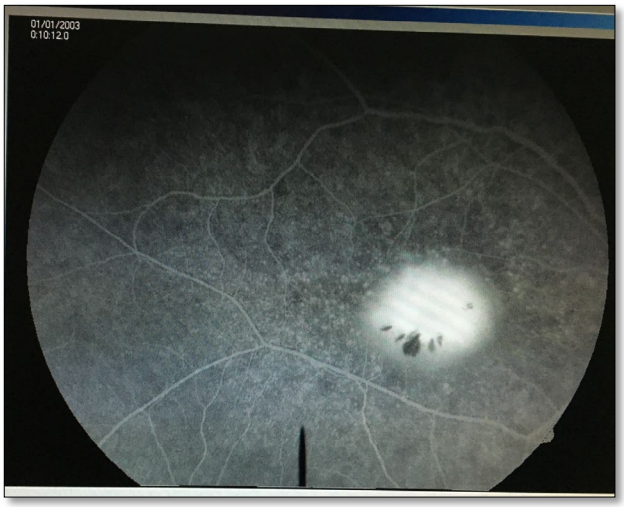

Figure 5. FA right eye showing hyper fluorescent lesion at macula with hypo pigmented spots inferiorly.

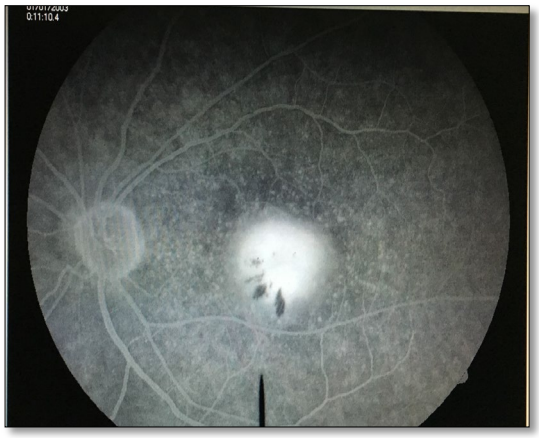

Figure 6. FA of left eye showing hyper fluorescent lesion at the macula with few hypo fluorescent spots inferiorly.

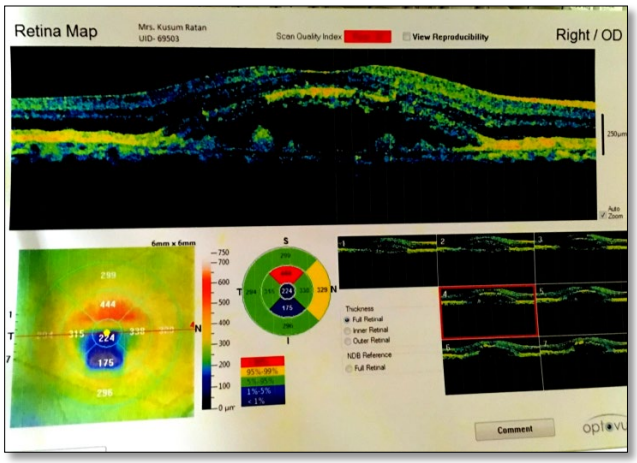

Figure 7. Right eye OCT showing hypo reflective region beneath the RPE.

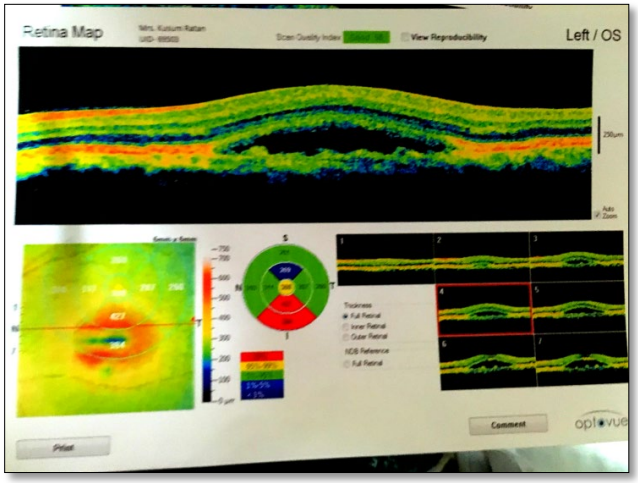

Figure 8. Left eye OCT showing hypo reflective region beneath the RPE.

DISCUSSION

The differential diagnosis of vitelliform dystrophy is central serous retinopathy, best's disease, inflammatory retinitis (toxoplasma retinalis), retinal pigment epithelial detachment, macular drusen and other macular degenerations.

This disease was an adult onset foveolar vitelliform dystrophy because:

1. Age of presentation was 60 years (>30 years).
2. Normal EOG findings.
3. Small lesion with the size of one-disc diameter at the macula.
4. Fluorescein angiography showed hyper fluorescent lesion in the macular region approximately the size of one-disc diameter.
5. The visual acuity is minimally subnormal.

The progression of this disease can lead to a slow decline in visual acuity, with metamorphopsia and red-green dyschromatopsia in the later stages. The visual acuity decline was found to be symmetrical and in some cases can be improved with a hyperopic correction, which could be due to slightly elevated macular lesions. The familial preponderance was also noted. The fluorescein angiography generally shows a hypo fluorescent fovea with a hyper fluoroscent ring around the macula. The OCT findings found a hyoreflective lesion at the level of RPE, showing a neurosensory detachment above the RPE suggestive of chroidal pathology. The sub retinal exudation is evidently not associated with a neovascularization or hemorrhages [2]. The electrical studies are useful in differentiating adult vitelliform dystrophies from true vitelliform dystrophy as Best's disease **(Table 1)**.

Table 1. Difference between true and adult vitelliform dystrophies.

Characteristics	True Vitelliform Dystrophy	Adult Vitelliform Dystrophy
Age on onset	<20 years	>40 years
Size of lesion	1-3 disc diameters	1/3-1 disc diameter
Symmetry	+	+++
Visual acuity	Normal to slightly subnormal	Initially 20/30-20/80
Color vision	Red-green anomaly in late stages	Red-green anomaly in late stages
Fluorescein angiography	Vitelliform lesion with hypo fluorescence	Vitelliform lesion with ring-like RPE defect
EOG	Sub-normal	Normal
Genetic aspects	Autosomal dominant	No definite pattern

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