

Adult-onset Foveomacular Vitelliform Dystrophy with Retinal Folds

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Background: Adult-onset foveomacular vitelliform dystrophy is characterized by a solitary, oval, slightly elevated, yellowish subretinal lesion of the fovea. We examined a patient with adult-onset foveomacular vitelliform dystrophy with stellate retinal folds by optical coherence tomography and scanning laser ophthalmoscopy.

Case: A 58-year-old Japanese woman with a complaint of decreased vision in her right eye was diagnosed as having adult-onset foveomacular vitelliform dystrophy.

Observations: Ophthalmoscopic examination revealed a yellowish lesion of one-third disc diameter in size at the fovea in the right eye. Fluorescein angiography demonstrated an irregular block of choroidal fluorescence corresponding to the yellowish lesion, which was surrounded by stellate retinal folds. Optical coherence tomography images showed a steep elevation of the retinal pigment epithelium (RPE) as a focally protruded reflective band over an optically clear space. Scanning laser ophthalmoscopy provided morphologic enhancement in the specifically affected layers of the macula. Using an argon green laser, band-shaped bright reflexes were seen in the right fovea. The helium-neon laser revealed a bright patch corresponding to the yellowish lesion over the fovea, which was surrounded by stellate retinal folds. The diode laser revealed a bright patch corresponding to the yellowish lesion.

Conclusion: The stellate retinal folds of this patient were considered to be caused by the steep elevation of the RPE with an extracellular accumulation of the vitelliform deposits.
Jpn J Ophthalmol 2001;45:533–537 © 2001 Japanese Ophthalmological Society

Key Words: Adult-onset foveomacular vitelliform dystrophy, optical coherence tomography, retinal fold, retinal pigment epithelium, scanning laser ophthalmoscopy.

Introduction

Adult-onset foveomacular vitelliform dystrophy,¹ also known as adult vitelliform macular dystrophy,^{2,3} foveomacular dystrophy-adult type,⁴ adult vitelliform macular degeneration,^{5,6} pseudovitelliform macular degeneration,⁷ and adult-onset foveomacular pigment epithelial dystrophy^{8,9} is characterized by a solitary, oval, slightly elevated yellowish subretinal lesion of the fovea.¹⁰ It appears ophthalmoscopically similar to the vitelliform or egg-yolk stage of Best's disease, but shows no inheritance pattern.¹¹ Initially the yellow lesion may be present in only one eye. The

size is usually one-third to one disc diameter, and small yellow flecks are seen in the paracentral lesion. Electro-oculographic testing reveals a normal or only slightly reduced Arden's ratio, which is intensely abnormal in Best's disease. The prognosis is optimistic, as most patients retain reading vision throughout life.

Occasionally this disease may be misdiagnosed as serous detachment of the retinal pigment epithelium (RPE). We describe a case of adult-onset foveomacular vitelliform dystrophy with stellate retinal folds, which was analyzed by optical coherence tomography (OCT) and scanning laser ophthalmoscopy (SLO).

Case Report

The patient was a 58-year-old Japanese woman with a complaint of decreased vision in her right eye for 6 months. She had been treated for open-angle

Received : December 20, 2000

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glaucoma with β -blocker for 10 years. Her visual acuity was 0.3 (n.c.) in the right eye, and 1.0 (n.c.) in the left eye. The intraocular pressure was 16 mm Hg in both eyes. Slit-lamp examination demonstrated no abnormality in the anterior segment of either eye. Ophthalmoscopic examination revealed a yellowish lesion of one-third disc diameter in size at the fovea in the right eye. In addition, stellate retinal folds were seen around the right fovea (Figure 1). In the left eye, yellowish small spots were arranged at the macula. Biomicroscopy revealed yellowish deposits that appeared to lie deep in the retina. The optic disc had glaucomatous cupping in both eyes. Fluorescein angiography (FAG) demonstrated an irregular block of choroidal fluorescence, corresponding to the yellowish lesion. In addition, stellate retinal folds surrounding the macular lesion were demonstrated (Figure 2).

SLO was applied to this patient. Using the argon blue laser, a dark oval area was seen in the fovea in both eyes. With the argon green laser, band-shaped areas of bright reflex were seen in the right fovea (Figure 3A). The helium-neon laser revealed intensely bright reflex corresponding to the yellowish lesion. In addition, stellate retinal folds were seen (Figure 3B). The diode laser revealed a foveal bright patch corresponding to the yellowish lesion, and stellate folds were clearly observed (Figure 3C). OCT images showed a thickening of the retina with reduced reflectivity in the macula. In addition, a small elevated area of the RPE was demonstrated as a focally elevated reflective band over an optically clear

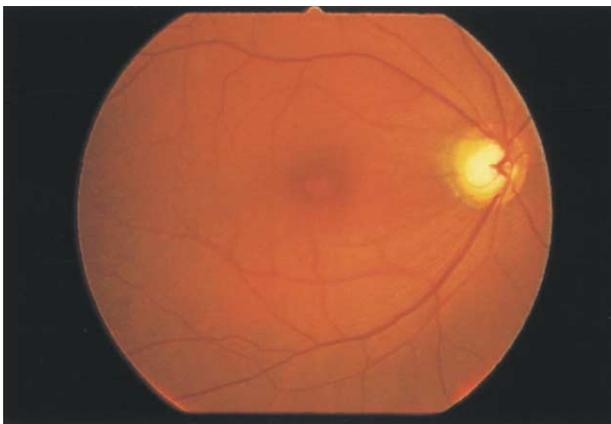


Figure 1. Fundus photograph of right eye of 58-year-old woman diagnosed as having adult-onset foveomacular vitelliform dystrophy. Yellowish lesion of one-third disc diameter in size surrounded by stellate retinal folds is seen in right fovea.

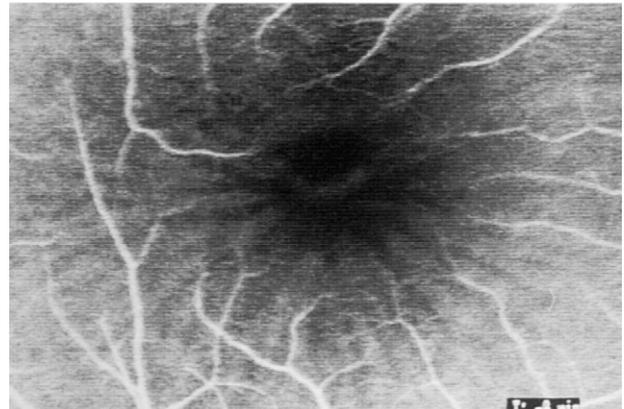


Figure 2. Fluorescein angiogram of right eye. Irregular block of choroidal fluorescence is seen in right fovea surrounded by stellate retinal folds.

space. The detached RPE is more highly reflective than normal, perhaps due to morphological changes in the RPE cells themselves. The angle of the RPE detachment was steep, probably because of the tight adherence of RPE cells to Bruch's membrane at the edge of the detachment (Figure 4). In the left eye, the foveal RPE layer was highly reflective compared to the perifoveal region.

The results of color vision testing with Ishihara's pseudochromatic plates were normal in both eyes. Automated perimetry (Octopus) revealed mildly depressed sensitivity in the pericentral visual field in both eyes. The scotopic electroretinogram with a single-white flash was normal in both eyes. The multifocal electroretinogram demonstrated subnormal cone responses in the affected foveal region bilaterally (Figure 5). The electro-oculogram (EOG) was normal with a light-to-dark ratio of 1.80 in both eyes.

Discussion

The differential diagnosis of vitelliform or egg yolk lesions should include Best's disease, adult-onset foveomacular vitelliform dystrophy, retinal pigment epithelial dystrophies such as reticular dystrophy or patterned dystrophy, central serous chorioretinopathy, inflammatory diseases such as toxoplasmosis, retinal pigment epithelial detachment, macular drusen, and age-related macular degeneration. Among these disorders, a diagnosis of Best's disease or adult-onset foveomacular vitelliform dystrophy should probably be based on FAG findings, which can demonstrate a block of choroidal fluorescence corresponding to the yellowish lesion.⁵ Electrical studies are useful in distinguishing adult-onset foveomacular vitelliform dystrophy from Best's disease.

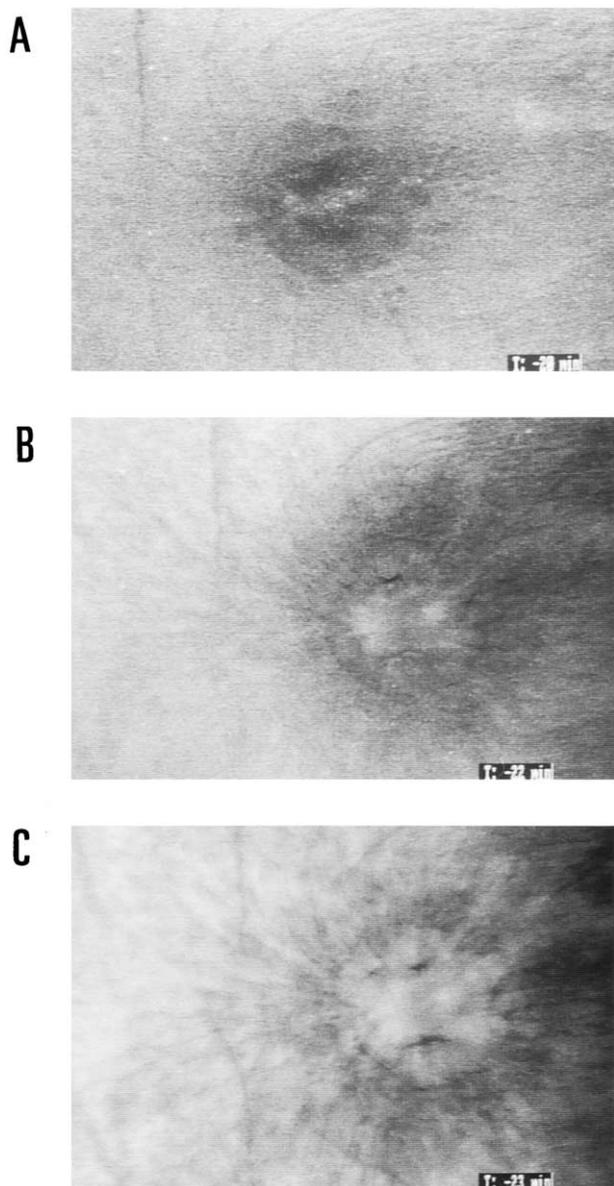


Figure 3. Images of right fundus by scanning laser ophthalmoscopy. (A) Argon green laser: Band-shaped bright reflex is seen over dark area of right fovea. (B) Helium-neon laser: Extremely bright patch is seen corresponding to oval distribution of yellowish deposits in right fovea. Retinal folds are also clearly visible. (C) Diode laser: Bright patch is seen in right fovea.

There was no markedly abnormal EOG, which is found in Best's disease, in our case.^{5,11} In our patient, the fundus abnormality, the reduced foveal response on multifocal electroretinogram and the normal EOG (Arden's ratio: 1.80) were compatible with a diagnosis of adult-onset foveomacular vitelliform dystrophy.

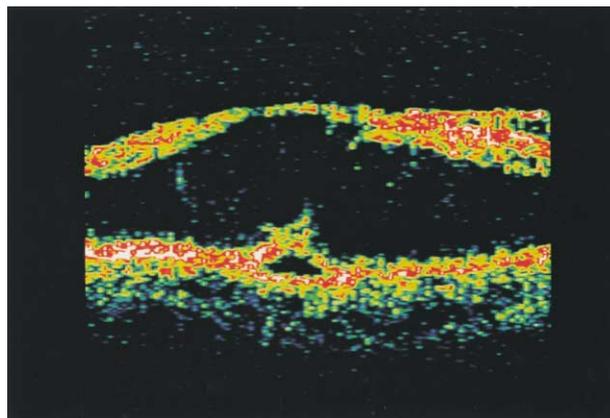


Figure 4. Optical coherence tomography image of right eye. Elevated area of retinal pigment epithelium (RPE) is demonstrated as focally elevated reflective band over optically clear space. Detached RPE is more highly reflective than normal.

The histopathologic study reported by Patrinely et al⁴ demonstrated massive amounts of lipofuscin within the abnormal pigment epithelial cells by ultraviolet fluorescent microscopy and transmission electron microscopy. Transmission electron microscopy confirmed the presence of myriad lipofuscin granules filling the cytoplasm of the retinal pigment epithelial cells and displacing a few melanosomes apically. Lipofuscin is generally acknowledged to be a yellowish wear-and-tear metabolic by-product that accumulates with age in the retinal pigment epithelium. The subpigment epithelial collagenous plaques were devoid of lipofuscin. Therefore, in adult-onset foveomacular vitelliform dystrophy, an abnormal accumulation of lipofuscin granules in the RPE can generate a yellowish subretinal deposition.^{4,11}

In adult-onset foveomacular vitelliform dystrophy, the ophthalmoscopic criteria have expanded to include a variety of sizes and patterns of yellow deposits in the fovea. The lesions may range in size from one-third to one disc diameter and may surround paracentral drusen. In older lesions, there may be both fading of yellow material and additional atrophic changes in the retinal pigment epithelium. In Best's disease, observation with monochromatic light was known to give a better picture of the typical cystic lesion. Krill et al¹² reported that studies with monochromatic light show localization of the typical early lesion at least as deep as the pigment epithelium. Deutman¹³ showed that panchromatic film, which has a maximum spectral sensitivity of 595 nm, gives a better picture of the typical cystic lesion than does orthochromatic film, which has a maximum spectral sensitivity of 580 nm.

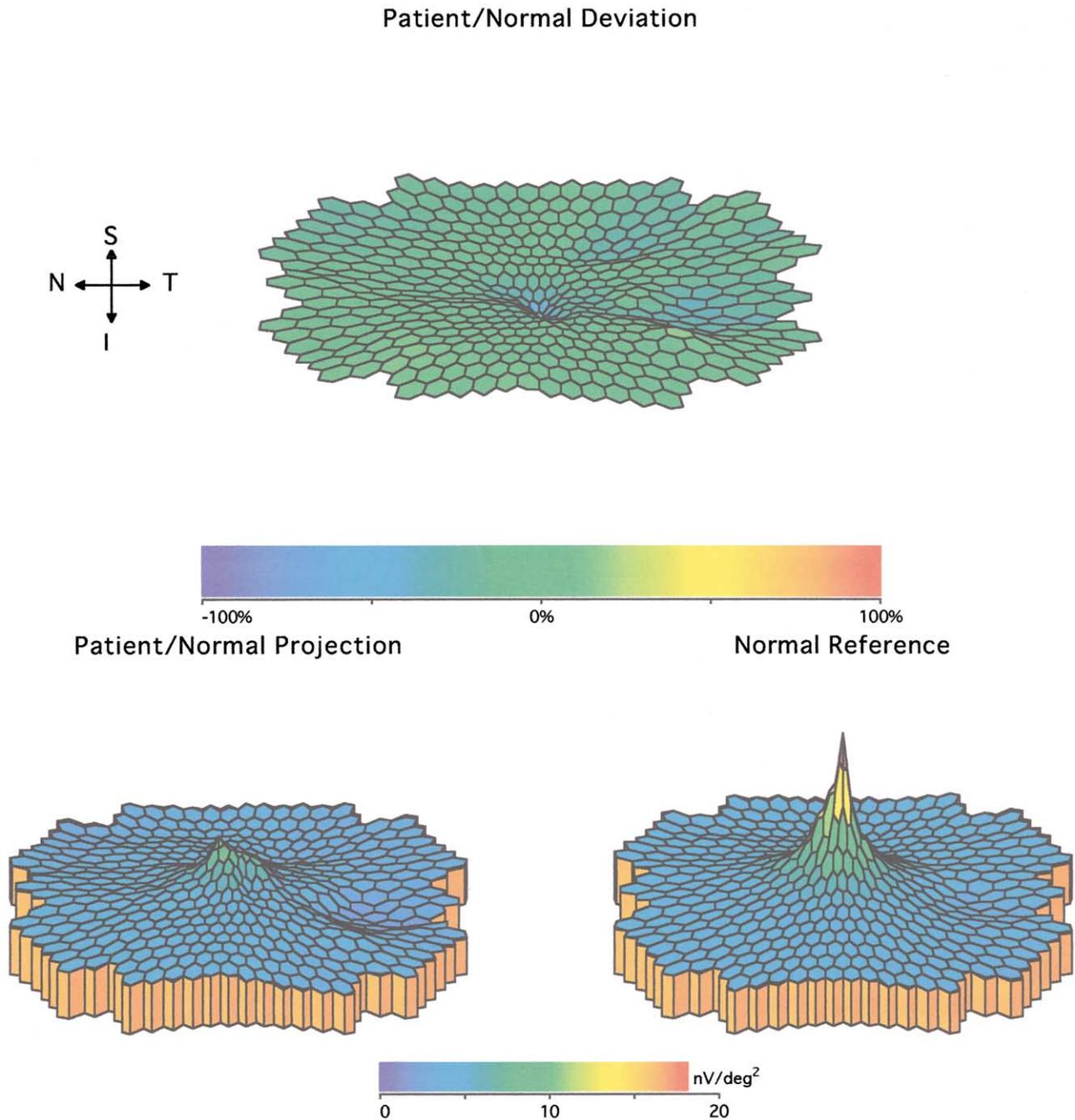


Figure 5. Multifocal electroretinogram of right eye. Subnormal cone responses in affected foveal region are seen.

The SLO from Rodenstock provides an image of the fundus by a scanning laser beam. Because of its confocal retinal scanning capabilities, with the SLO, which uses argon blue at 488 nm, argon green at 514 nm, helium-neon at 633 nm, and diode at 780 nm, it is possible to achieve highly resolute imaging with good contrast.^{14,15} In our case, SLO provided a morphologic

enhancement that correlated with known and predictable abnormalities in specifically affected layers of the macula in the adult-onset foveomacular vitelliform dystrophy. The discrepancy between the argon green and helium-neon laser proved that the yellowish material existed in the outer retina. In addition, the view with the helium-neon laser suggested a structural ab-

normality in the retinal pigment epithelium layer at the fovea. Furthermore, in this case, the diode laser revealed a bright patch in the fovea. This suggested an abnormality in the deeper structure of the fundus such as Bruch's membrane. Because we have not seen such findings in the ocular fundus of normal volunteers, these changes in the deeper retina seemed to be pathognomonic for adult-onset foveomacular vitelliform dystrophy.

For the study of hereditary retinal degeneration, OCT has provided an in vivo alternative to histopathologic pictures.^{16,17} In our patient, the foveal RPE layer was highly reflective compared to the perifoveal region. This is compatible with the known histopathological finding of adult-onset foveomacular vitelliform dystrophy, indicating an abnormal accumulation of lipofuscin granules in the RPE.¹⁸ In the right fovea, OCT images presented a thickening of the neurosensory retina with reduced reflectivity. In addition, an elevated area of the RPE was demonstrated as a focally elevated reflective band over an optically clear space in the right eye. These findings demonstrated the formation of closed cystic spaces with abnormal yellowish substances in the sub-RPE space. The angle of the RPE detachment was steep, because of the tight adherence of RPE cells to Bruch's membrane at the edge of the detachment, resulting in stellate retinal folds. OCT in conjunction with SLO was useful in the diagnosis of the foveal lesion in this patient. Practitioners should consider adult-onset foveomacular vitelliform dystrophy when making a differential diagnosis of stellate macular folds.

References

1. Battaglia Parodi M, Iustulin D, Russo D, Ravalico G. Adult-onset foveomacular vitelliform dystrophy and indocyanine green videoangiography. *Graefes Arch Clin Exp Ophthalmol* 1996;234:208-11.
2. Brecher R, Bird AC. Adult vitelliform macular dystrophy. *Eye* 1990;4:210-5.
3. Felbor U, Schilling H, Weber BH. Adult vitelliform macular dystrophy is frequently associated with mutations in the peripherin/RDS gene. *Hum Mutat* 1997;10:301-9.
4. Patrinely JR, Lewis RA, Font RL. Foveomacular vitelliform dystrophy, adult type: a clinicopathologic study including microscopic observations. *Ophthalmology* 1985;92:1712-8.
5. Epstein GA, Rabb MF. Adult vitelliform macular degeneration: diagnosis and natural history. *Br J Ophthalmol* 1980;64:733-40.
6. Bloom LH, Swanson DE, Bird AC. Adult vitelliform macular degeneration. *Br J Ophthalmol* 1981;65:800-1.
7. Fishman GA, Trimble S, Raab MF, Fishman M. Pseudovitelliform macular degeneration. *Arch Ophthalmol* 1977;95:73-6.
8. Vine AK, Schats H. Adult-onset foveomacular pigment epithelial dystrophy. *Am J Ophthalmol* 1980;89:680-91.
9. Dubovy SR, Hairston RJ, Schats H, et al. Adult-onset foveomacular pigment epithelial dystrophy: clinicopathologic correlation of three cases. *Retina* 2000;20:638-49.
10. Gass JDM. A clinicopathologic study of a peculiar foveomacular dystrophy. *Trans Am Ophthalmol Soc* 1974;72:139-56.
11. Gass JDM. Best's disease. In Gass JDM, ed. *Stereoscopic atlas of macular disease*, St. Louis: CV Mosby, 1987:246-51.
12. Krill AE, Morse PA, Potts AM, Klein BA. Hereditary vitelliform macular degeneration. *Am J Ophthalmol* 1966;61:1405-15.
13. Deutman AF. *The hereditary dystrophies of the posterior pole of the eye*. Springfield: Charles C Thomas, 1971:198-299.
14. Manivannan A, Kirkpatrick NJP, Sharp PF, et al. Clinical investigation of an infrared digital scanning laser ophthalmoscope. *Br J Ophthalmol* 1994;78:84-90.
15. Woon WH, Fitzke FW, Bird AC, et al. Confocal imaging of the fundus using a scanning laser ophthalmoscope. *Br J Ophthalmol* 1992;76:470-4.
16. Hee MR, Izatt JA, Swanson EA, et al. Optical coherence tomography of the human retina. *Arch Ophthalmol* 1995;113:325-32.
17. Puliafito CA, Hee MR, Lin CP, et al. Imaging of macular diseases with optical coherence tomography (OCT). *Ophthalmology* 1995;102:217-29.
18. Puliafito CA, Hee MR, Schuman JS, Fujimoto JG. Retinal dystrophies. In: *Optical coherence tomography of ocular disease*. Thorofare: SLACK, 1996:269-75.