Seven-year longitudinal multimodal imaging analysis of a patient with evolving adult onset foveomacular vitelliform dystrophy

Abstract: We present the natural progression of an evolving case of adult onset foveomacular vitelliform dystrophy (AOFVD) over a seven-year period, emphasizing the role of multimodal imaging in the diagnosis, staging and management of this condition.

I. Case History
- **Demographics**: 79-year-old Caucasian male followed since 2011 with AOFVD OS > OD since 2014
- **Chief Complaint**: Patient reports stable vision right eye and “wavy” vision left eye when watching TV
- **Ocular History**: Glaucoma suspect secondary to asymmetric cupping (OS>OD), Pseudophakia s/p CE/PCIOL OU, Melanoma removal LUL 1972, no recurrence or metastasis
- **Medical History**: Anxiety, Diabetes mellitus type II, Diverticulosis, Hyperlipidemia, Hypertension, Melanoma, Polyp of colon, Sciatica
- **Medications**: Amlodipine 10mg, Atenolol 50mg, Atorvastatin calcium 40mg, Fosinopril 20mg, Glipizide 10mg, Hydrochlorothiazide 25mg, Meloxicam 15mg, Metformin HCL 500mg, Multivitamin/Opt Nation/Lutein cap 1 tablet P.O. BID, Pioglitazone HCL 30mg, Tamsulosin HCL 0.4mg
- **Other salient information**: Patient underwent retina consult to confirm diagnosis of AOFVD in 2014

II. Pertinent findings
- **Clinical**:
  - BCVA OD 20/20, OS 20/30
  - Ancillary Tests: normal confrontation fields / pupils / EOMs;
    - Amsler grid: WNL OD, metamorphopsia superior to fixation OS
  - Slit lamp Exam: unremarkable except for mild blepharitis OU and PCIOL OU; IOP 10mmHg/11mmHg
  - Posterior segment:
    - Optic nerves: small cupping with pink/healthy rim tissue 360 and distinct/flat margins OU
    - Macula
      - OD: pigment mottling throughout fovea with scattered pinpoint, hard drusen-like deposits, (-) CNVM
      - OS: central, large round yellow elevated lesion with surrounding pigmentary changes, few scattered pinpoint, hard drusen-like deposits, (-) CNVM
- **Imaging/Ancillary testing**: Extensive multimodal imaging including spectral domain optical coherence tomography (SD-OCT), fundus autofluorescence (FAF), infra-red (IR), and fundus photography were performed routinely dating back to 2011. As the condition evolved, sequential multimodal imaging unveiled the diagnosis of AOFVD with a central, round yellow vitelliform lesion between the retinal pigment epithelium (RPE) and photoreceptors (PRs). The various stages of AOFVD (stage 1: vitelliform, stage 2: pseudohypopyon, and stage 3: vitelliruptive), and imaging features will also be illustrated and discussed.

III. Differential diagnosis
- Primary/leading: Stage 2/3 AOFVD (pseudohypopyon with vitelliruptive characteristics) OS>OD
- Others: Non-exudative age-related macular degeneration OU, Best disease OU, Central serous chorioretinopathy, Large confluent soft drusen, Pattern dystrophy

IV. Diagnosis and discussion
  AOFVD was first described by Gass in 1974 as an uncommon heterogeneous macular dystrophy with varying appearance, and in the early stages can resemble a drusenoid PED or dry ARMD. It shows a variable inheritance pattern, from autosomal dominant to sporadic, and is thought to stem from mutations of the BEST1 or photoreceptor peripherin gene. Age of onset is around 4th to 6th decade, and the condition is often diagnosed incidentally as patients are generally asymptomatic, or mildly symptomatic with central/paracentral blur and mild metamorphopsia. AOFVD typically presents as a bilateral but asymmetric yellow macular lesions with variable size, shape and pigmentation. The advent of multimodal imaging has enhanced our understanding of AOFVD and aids in the diagnosis and management of this condition.
Characterized as part of a larger group known as Acquired Vitelliform Lesions (AVLs), AOFVD lesions represent accumulation of lipofuscin and melanosome waste material between the RPE and photoreceptor layer. Permanent vision loss from AVLs generally results from foveal atrophy or choroidal neovascular membrane (CNVM). SD-OCT is a useful tool for identifying structural features and assists in staging and monitoring disease for progression. Newly introduced OCT-angiography has been studied to show that patients with AOFVD exhibit a vascular network rarefaction with less blood vessels at the superficial and deep capillary plexuses and the choriocapillaris layer. OCT-angiography may be able to detect CNVM formation in these patients earlier than conventional SD-OCT, fluorescein angiography or ICGA imaging modalities.

FAF and IR are other useful tools for clinical interpretation. FAF in a patient with AOFVD can be uniform (matching the appearance of an eye without AOFVD), diffusely patchy in appearance, or have a focal central autofluorescent circular lesion. IR imaging is excellent at highlighting hyper-reflective retinal areas.

Similar to Best disease, AOFVD can be characterized by stages, though likely asymmetric between eyes. Multimodal imaging greatly aids in properly staging and following the course of disease progression. AOFVD staging subsequently includes: stage 1 vitelliform, stage 2 pseudohypopyon, stage 3 vitelliruptive and stage 4 atrophic. Our case demonstrates the change and progression of AOFVD over the course of seven years depicted by SD-OCT, FAF, infrared images and fundus photos. The waxing and waning nature of the disease course is clearly illustrated in addition to the variability of presentation and various staging.

V. Treatment, management
- Repeat DFE + multimodal imaging every 6 months
- Home Amsler grid for self-monitoring
- Consider UV protection and tinting for glare reduction and contrast enhancement
- Retina consult if CNVM is suspected
- Low vision evaluation if warranted

VI. Conclusion
This phenomenal case of AOFVD with seven-years of follow-up, assisted by the use of advanced multimodal imaging, furthers our understanding of the natural progression and pathophysiological characteristics of this condition. This case contributes to our understanding of AVLs, and highlights the importance of advanced imaging in the care of our patients. It serves to broaden our differential diagnoses when encountering ARMD-like cases, and empowers optometrists to play an integral role in the diagnosis and management of patients with AOFVD.

VII. References


