Adult-onset foveomacular vitelliform dystrophy: a unique presentation

Abstract:
A case of adult-onset foveomacular vitelliform dystrophy with an unusual noncentral vitelliform lesion. This variant is clinically indistinguishable from the cuticular drusen subtype of age-related macular degeneration at this stage in the disease.

1. Case history
   a. 73 y.o. white male
   b. Chief complaint: blurry vision OU (longstanding, constant, no relief, stable)
      i. Pt reports that vision began deteriorating 10-15 years ago, eye exams before that time did not have positive findings per the patient
   c. Ocular/medical history
      i. Last eye exam: 1 year ago at VA SLC in optometry department (diagnosed with ARMD, prescribed AREDS 2 supplements BID)
         1. Evaluated several years ago by unknown retinal specialist outside VA system, but was not treated at that time
      ii. Medical conditions: hypertension, hypothyroidism, vitamin D insufficiency, hyperlipidemia, osteopenia, aortic stenosis
   d. No ocular family history
   e. Medications: atorvastatin, vitamin D/calcium supplement, cholecalciferol, hydrochlorothiazide, levothyroxine, metoprolol, AREDS 2, spironolactone, terazosin, aspirin, multivitamin

2. Pertinent findings
   a. Best corrected visual acuity
      i. OD: 20/100+
      ii. OS: 20/70-
   b. Normal CVF, pupils, EOM’s
   c. Anterior segment unremarkable OU except:
      i. 1+ cortical cataract and 1+ nuclear cataract OU
   d. Goldmann applanation OD, OS
      i. 17, 17 @ 9:47 AM
   e. Posterior segment unremarkable OU except:
      i. PVD OU
      ii. Well defined central retinal atrophy OU (fundus photos taken)
         1. Subretinal fluid superior temporal to atrophy OD
         2. 2.5DD x 1DD oval elevated vitelliform lesion temporal to macula OS
         3. Scattered hard drusen along arcades (nasal>temporal) especially near optic disc and in periphery OU
         4. Findings confirmed with OCT

3. Differential diagnosis
   a. Primary: Adult-onset foveomacular vitelliform dystrophy
      i. Atrophic stage OU, w/ atypical non-central lesion
   b. Secondary: Age-related macular degeneration, geographic atrophy OU
      i. Cuticular drusen subtype w/ associated vitelliform lesion
c. Bestrophinopathy OU

d. Best’s disease OU (atrophic stage)

4. Diagnosis and Discussion

a. Atypical end stage adult-onset foveomacular vitelliform dystrophy (AFVD) OU
   i. Unusual multifocal presentation consistent with atrophic vitelliform dystrophy, has been associated with scattered drusen

b. ARMD w/ cuticular drusen is a very high differential diagnosis
   i. Can result in vitelliform lesions, however drusen typically present at a younger age (it remains debatable as to whether the patient’s previous providers would have told him this information had it been present)
   ii. Clinically, it is very difficult to differentiate between this form of ARMD and AFVD

c. Best’s disease is unlikely as patient was not diagnosed with any eye disease prior to the age of 50 and the presence of scattered drusen in the fundus negates the diagnosis

d. Bestrophinopathy
   i. Extremely rare autosomal recessive condition usually with more scattered vitelliform lesions

5. Treatment/Management

a. Referral to retinal specialist for evaluation of subretinal fluid and confirmation of diagnosis. Discussed low vision options. Continue AREDS 2 BID until diagnosis is established.
   i. Pt scheduled with retina specialist at VA SLC 9/5/17

b. Brief literature review:

6. Conclusion

a. Though in this case it could be, it’s not always ARMD!
   i. This “classic” case of an older white male with central vision loss and corresponding macular atrophy should lead us to think of ARMD initially, but rule it out when the findings do not fit. AREDS 2 in this case may not be helpful and the burden of taking the vitamin could have been avoided. (Granted this patient received AREDS 2 through the VA).

b. It’s not rare if it’s in your chair.
   i. It’s not everyday something strange will walk into your office, but when things don’t add up the way you expect them to, it’s nice to have a few other differentials to turn to.