Atypical new diagnosis of X-linked bilateral macular retinoschisis in a 67-year-old Vietnam combat veteran

Case Summary

1. Case History
   a. Demographics/Chief complaint
      i. 67 yo Caucasian Male presents for diabetic eye exam with no ocular complaints
   b. Ocular/Medical History
      i. Ocular History: DM2 w/ no retinopathy and Refractive error w/ presbyopia at LEE (10/2014) w/ BCVA 20/20 OD, OS
         1. Hx of LASIK 20+ years ago, pre-lasik refractive error unknown
      ii. Medical History
         1. DM2, Afib, HTN, Hyperlipidemia, Hypothyroid, BPH
      iii. Medications
         1. Metformin, Warfarin, Lisinopril, Glipizide, Finasteride, Levothyroxine, Propanolol, Tamulosin

2. Pertinent findings
   a. Clinical
      i. BCVA: 20/20-1 OD, 20/25+2 OS
      ii. Anterior Segment: Mild NS, otherwise WNL OU
      iii. IOP: 16/16 Applanation
      iv. Posterior segment
         1. Subtle tractional star like appearance at fovea OD, OS
         2. Single CWS inferior arcade OS
   b. Imaging
      i. OCT – schisis separation at fovea OD, OS
      ii. Red-free photos – obvious spoke like pattern at fovea OD, OS

3. Differential Diagnosis
   a. Myopia induced macular retinoschisis
   b. Goldmann Farve syndrome
   c. Juvenile X-linked macular retinoschisis
   d. Optic pit maculopathy
   e. Degenerative retinoschisis

4. Diagnosis and Discussion
   a. Juvenile X-linked bilateral macular retinoschisis
   b. Bilateral splitting of superficial retina
      i. Always macular schisis, 50% associated peripheral schisis
   c. Genetic disease affects almost exclusively males
   d. Complete penetrance, variable expressivity
e. Mutation in RS1 at XP22
   i. Found bipolar cells and photoreceptors, useful in adhesion
f. Characteristic features: spoke like appearance at fovea, schisis separation on OCT
g. VA varies 20/20-20/200
   i. Stabilizes 2\textsuperscript{nd}-4\textsuperscript{th} decade and then progresses due to RPE atrophy
h. Complications
   i. Peripheral schisis, Retinal detachment, vitreous hemorrhage
i. Diagnostic imaging
   i. Spectral domain OCT
   ii. Red Free photographs
   iii. Multifocal ERG

5. Treatment and Management
   a. Monitor if no secondary complications
   b. Genetic testing/counseling
   c. Educational support for children
   d. Treat secondary complications w/ surgical intervention
   e. Avoid high risk activities
   f. Regular follow up – 6 mos to 1 year
   g. Future treatment options may include
      i. Topical dorzolamide
      ii. Gene therapy –

6. Conclusion
   a. Varying acuity, diagnosis can be missed
   b. Use of appropriate diagnostic tools is important in diagnosis
   c. Genetic testing/Counseling plays a key role in management
   d. New potential therapies could change treatment options drastically

7. Bibliography