Splitting of Differentials: a New Type of Foveal Retinoschisis

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Abstract

Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis (SNIFR) is a newly described entity that lacks a known hereditary or acquired predisposition. This case highlights the clinical characteristics, differential diagnoses, treatment, and management of this novel diagnosis.

I. Case History

A. 73 year old white male presents complaining of distance vision blur with current specs

B. Ocular History
   a. Follow-up for foveal retinoschisis OD
   b. Stable choroidal nevus OD with overlying drusen, (-)subretinal fluid
   c. Dry Eye Syndrome secondary to meibomian gland dysfunction and anterior blepharitis
   d. Mild Cataracts, OU; not visually significant
   e. Compound hyperopic astigmatism OU; Presbyopia OU

C. Medical History
   a. Hypertension
   b. Hyperlipidemia
   c. Obstructive sleep apnea
   d. Tachycardia
   e. PTSD

D. Medications
   a. Amlodipine 5mg
   b. Metoprolol 25mg
   c. Atorvastatin 20mg
   d. Sertaline 200mg
   e. Trazodone 300mg
   f. Clonezapam 0.5mg
   g. Aspirin 81mg

E. No family history of eye diseases
F. No family history of systemic disease

II. Pertinent findings

A. Clinical Findings
   a. Entrance Testing:
      • PERRL (-)APD
Motilities: Full OU
CVF: FTFC OD/OS
BCVA: 20/20-1 OU

b. Anterior Segment
i. Pingueculae temporal OU, trace injection OU
c. Tonometry
i. 18 OD, 17 OS @ 1:42 PM (GAT)
d. Posterior Segment
i. Lens: Trace NS OU
ii. C/D: 0.35R OD, 0.4V/0.35H OS; Healthy rim tissue OU
iii. Macula:
   1. OD: Faint stellate appearance inferior fovea
   2. OS: Clear and flat
iv. Posterior Pole:
   1. OD: 2DD choroidal nevus with overlying drusen 1 DD nasal to ONH without elevation
   2. OS: WNL
v. Vessels: Mild A/V crossing changes
vi. Periphery: Flat and intact, no holes, tears, or abnormalities 360 OU

B. Imaging
a. Macular OCT:
   i. OD: Intraretinal splitting at the outer plexiform layer at both the fovea and temporal to the fovea; photoreceptor integrity line intact (-)subretinal fluid
   ii. OS: WNL, photoreceptor integrity line intact (-)subretinal fluid
b. Fundus Photography
   i. OD: 2DD choroidal nevus 1DD nasal to ONH; trace stellate appearance at fovea (-)edema
   ii. OS: WNL, macula clear, (-)edema

C. Ancillary Testing
a. Amsler Grid: No metamorphopsia or scotomas noted OU

III. Differential diagnosis

A. Primary: Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis (SNIFR) (Ober et al. 2014)

B. Others
   a. X-linked juvenile retinoschisis
   b. Degenerative Retinoschisis
   c. Enhanced S-cone dystrophy (Goldmann-Favre Syndrome)
   d. Autosomal dominant retinoschisis
   e. Neovascular AMD

C. Potential Causes of Retinoschisis
   a. Myopic Degeneration
   b. Optic Pit Maculopathy
   c. Glaucoma
d. Myotonic Dystrophy

IV. Diagnosis and discussion

A. SNIFR is a recently classified entity (Ober et. al. 2014) with several identifying characteristics. Clinically, a subtle radial spoking will be visible at the fovea, giving the stellate appearance. Patients also generally demonstrate a splitting at the outer plexiform layer with relatively preserved acuity (>20/40). A genetic linkage is unlikely as no family history of the RS1 mutation was found in any of the study patients. RS1 codes for retinoschisin, a secreted protein that promotes cell adhesion and cell to cell interactions. RS1 mutation has been found to be the disease causing gene in X-linked retinoschisis. Additionally, the condition was more likely to occur in females in the cited study, which is dissimilar to X-linked Retinoschisis, an almost exclusively male condition. The majority of patients had a unilateral presentation; however there were subjects with bilateral presentation. Further criteria include a lack of predisposing conditions such as the differential diagnoses listed above.

B. Diagnosis of Exclusion: Rule out differentials

i. X-linked Retinoschisis: Occurs early in life (school age children) with a bilateral presentation and a positive family history. Visual acuity deteriorates to about 20/100 (Molday et. al. 2012). Our patient is in his eighth decade of life and presented unilaterally with a BCVA 20/20 in the involved eye.

ii. Degenerative retinoschisis: Isolated case reports present foveal retinoschisis associated with degenerative retinoschisis (Han et. al. 1988). Our patient had a complete DFE with no peripheral retinoschisis noted.

iii. Enhanced S Cone dystrophy (Goldmann-Favre Syndrome): This condition is characterized by degenerative vitreal changes, cystoid maculopathy, retinoschisis in the posterior pole and/or the periphery, night blindness, and impaired visual acuity. (Theodossiadis et. al. 2000). The presentation of our patient, beyond the foveal retinoschisis, did not match.

iv. Autosomal Dominant Retinoschisis: This is another genetic variant with AD penetrance (Yassur et. al. 1982). Our patient had no family history of retinoschisis to his knowledge.

v. Neovascular AMD: Exudative AMD has the potential to create intraretinal fluid and cystic spaces from a choroidal neovascular membrane that may split the retina. Our patient had no clinical signs of AMD.

C. Rule out other potential causes of retinoschisis

i. Myopic Degeneration: Schisis-like degeneration occurs in highly myopic eyes in the presence of posterior staphylomas (Johnson 2012). Our patient was hyperopic and did not have a posterior staphyloma.

ii. Optic pit maculopathy: Schisis-like fluid builds up and a potential for retinal detachments occurs in the presence of optic pits. (Imamura et. al. 2010). Our patient did not have an optic pit.

iii. Glaucoma: Various case reports have shown a possible link between glaucomatous optic neuropathy and macular retinoschisis (Zhao and Li...
iv. Myotonic Dystrophy: A familial disease characterized by muscle wasting with ocular findings of reticular macular dystrophy, peripheral pigmentary retinopathy, ptosis, cataracts and in the recent cited case report, foveal schisis (Krishnan and Lochhead 2010). Our patient has no history of myotonic dystrophy or any signs of muscle weakness as per a primary care records review.

D. Based on clinical appearance, history, demographics, and exclusion of differentials, a diagnosis of the recently identified SNIFR was made.

V. Treatment, management

A. Treatment of SNIFR is commonly observational due to exceptionally good BCVA. Patients should be regularly followed with macular OCT’s to assess retinal architecture. Visual fields may also be done to assess loss of visual function, although it is not critical for diagnostic purposes.

B. For patients with concomitant macular degeneration, care should be taken to differentiate between stable intraretinal fluid vs. new, evolving intraretinal fluid due to neovascular AMD. (Casalino et. al. 2016)

C. Cases of X-linked retinoschisis have been treated with topical dorzolomide with limited success. Studies found a significant decrease in central macular thickness, with some improvement in VA. Drops may need to be taken up to five months before any appreciable change takes place. (Genead et. al. 2010). Caution must be taken in extrapolating the findings of this study to that of SNIFR. With etiological differences in onset between SNIFR and X-linked retinoschisis, studies must be done to evaluate the efficacy of topical dorzolamide on SNIFR.

D. Vitrectomy with a gas tamponade has been studied in response to foveal retinoschisis due to myopic maculopathy. Following surgery, the majority of eyes displayed eventual foveal re-attachment and an improvement in final visual acuity (Kobayashi and Kishi 2003). Again, extrapolation of these findings to SNIFR is problematic due to differences in pathophysiology.

E. Our patient is currently being monitored on a bi-annual basis since the onset of the condition. He continues to use the home amsler grid and has instructions to return if any changes should occur. The retina specialist that sees this patient in conjunction states that there is neither need nor likely benefit to attempt to re-attach the fovea when BCVA is 20/20.-.

VI. Conclusion

A. SNIFR is a rare, recently identified entity with very little research currently available. It is an important differential when assessing a foveal retinoschisis especially when the condition is unilateral and BCVA is exceptionally good. Due to the condition’s recent identification, there is no established treatment or management protocol. Additionally,
the fact that SNIFR is a nascent entity makes it difficult to assess what will happen with our patient in the future. Will the best corrected visual acuity remain the same or will it decline due to eventual atrophy? Will the retinoschisis self-resolve? Will unilateral cases become bilateral with time? Without answers to these questions, it is important for the clinician to follow-up with these patients on a consistent basis.

VII. References


