Treatment and Management of Juvenile X-linked Retinoschisis in a Hospital Setting:  
A Case Report

Andy T. Cheng, O.D., Cristina Llerena Law, O.D., F.A.A.O., Kristen Krummenacker, O.D.,  
Marilyn Vricella, O.D., Danielle Crane, O.D.

Abstract: A 6-year-old Arabic male being followed closely for anisometropic amblyopia  
presented for a refractive follow-up with a sudden decrease in visual acuity. New-onset  
progressive macular pathology was detected during a follow up visit. Assessment,  
electrodiagnostic testing, differential diagnoses, and treatment/management considerations are  
discussed.

I. Case History  
a. 6-year-old Arabic male  
b. No specific visual complaints from mom or patient  
c. Presence of refractive amblyogenic factor secondary to unequal hyperopic  
refractive errors OD>OS  
d. No significant medical history: full-term birth, (-) complications with  
pregnancy/birth  
e. No current medications  
f. (-) family hx of eye disease

II. Pertinent Findings  
a. Current VA (Snellen): OD 20/40, OS 20/50 (Note: previous measures of visual  
acuity with Allen picture matching indicated equal vision of 20/30 in each eye,  
which is the highest resolution on said chart)  
b. R/G color deficiency OD/OS  
c. While routine dilated and undilated assessments of posterior segment health  
conducted at three month intervals revealed no abnormalities, an abrupt loss of  
normal foveal reflex with RPE mottling and elevated, “spoke-wheel” macular  
appearance was noted during a routine refractive follow up visit  
d. (-) peripheral retina pathology  
e. OCT testing: Cystic spaces in the foveal and perifoveal regions OS>OD, located  
in the outer plexiform layer of the retina  
f. ERG testing revealed findings consistent with Juvenile Retinoschisis vs Atypical  
Retinitis Pigmentosa

III. Differential Diagnoses  
a. Leading: Juvenile X-linked Retinoschisis  
b. Stargardts Macular Dystrophy  
c. Best’s Disease  
d. Rod/Cone Dystrophy  
e. Atypical Retinitis Pigmentosa

IV. Case Discussion  
a. This patient initially presented to our clinic in May 2010 at 4 years old and was  
being followed at three-month intervals for refractive follow-ups and yearly for  
comprehensive eye exams to monitor for progression of amblyopia. At each of his  
visits, his visual acuity remained stable at the maximum of 20/30 OD/OS using
Allen picture matching and fundus exam showed no pathology. In June 2011, the patient’s vision was tested for the first time using Snellen visual acuity measure and his best corrected visual acuity was measured to be 20/30 OD/OS. Color vision testing done at this exam suggested a R/G color deficiency. Undilated assessment of the posterior pole showed no pathology and a positive foveal reflex at the macula. The patient returned in two weeks for a cycloplegic exam with dilation and a sudden loss of the foveal reflex OD/OS with associated RPE mottling was detected. Retinal elevation with a “spoke-wheel” type appearance typical of juvenile X-linked retinoschisis was also observed. These macular findings coincided with a more significant decrease in visual acuity in the left eye relative to the right eye. OCT testing revealed cystic spaces in the foveal and perifoveal region of the macula OU. The patient was referred to Bellevue Hospital for an electroretinogram that confirmed our leading differential diagnosis.

b. Other ocular signs that can be associated with juvenile X-linked retinoschisis
   i. Peripheral retinoschisis, typically inferotemporally
   ii. Vitreous hemorrhage
   iii. Severe atrophic changes at the macula
   iv. Mizuo phenomenon, a color change in the retina to a diffuse yellow or gray color with the onset of light after dark adaptation

V. Treatment & Management
   a. Close monitoring and patient education regarding other ocular complications that can be associated (i.e. vitreous hemorrhage, retinal detachment)
   b. Low vision aids such as large print textbooks, high contrast handouts and preferential seating in the classroom
   c. Genetic counseling

VI. Conclusion
   a. Clinical pearl: This case alerts each clinician of the importance of monitoring both ocular health AND refractive status concurrently when following patients with amblyogenic factors which could account for decreased vision. The importance of frequent visual function and refractive follow-ups for these patients is stressed.

VII. Bibliography