Bilateral foveal retinoschisis accompanying unilateral peripheral retinoschisis

Nilufer Kocak, Taylan A Ozturk1, Suleyman Kaynak

X-linked juvenile retinoschisis is a rare hereditary retinal disease characterized by a tangential splitting of the neurosensory retina which may cause early-onset visual impairment. Existence of the retinal neurosensory layer splitting on cross-sectional images of optical coherence tomography (OCT) and the absence of leakage on fluorescein angiography (FA) help confirming the diagnosis. Such diagnostic tests are also helpful in determining the management of the disease. However, most of the retinoschisis cavities remain stable and rarely extend to the posterior pole, many authors suggest laser prophylaxis to avoid the potential risk of retinal detachment due to holes in the outer retinal layer. Herein, we report a case with bilateral foveal retinoschisis accompanying unilateral peripheral retinoschisis who was evaluated with detailed ophtalmologic examination. Visual acuity, fundoscopy, OCT, and FA remained stable in the second year of follow-up after prophylactic argon laser treatment.

Key words: Fluorescein angiography, optical coherence tomography, prophylactic argon laser treatment, retinoschisis

Visual deterioration that progresses during the first to second decades of life is the most common symptom of retinoschisis, while amblyopia, strabismus, and nystagmus related with retinal detachment and vitreous hemorrhage may also be seen.1,2 The main clinical feature is foveal schisis with cystoid spaces and fine radial striae located in the macula. Peripheral retinoschisis occurs in approximately 50% of affected male patients. Vitreous hemorrhage, retinal detachment, and neovascular glaucoma may also be seen.3,4 Although there is no proven therapeutic approach for this disease, laser photocoagulation, cryotherapy, scleral buckle, and pars plana vitrectomy are considered as the main treatment options for retinoschisis especially complicated with vitreous hemorrhage or retinal detachment.4,8

Case Report

A 11-year-old boy was referred with complaints of bilateral blurred vision. The best corrected visual acuities were 10/20 in the right eye and 20/200 in the fellow eye. Anterior segment examination with slit lamp biomicroscopy was unremarkable. Funduscopy examination via indirect ophthalmoscopy revealed minimal macular edema in the right eye and diffuse pigmented changes as well as peripheral retinoschisis in the fellow eye [Fig. 1]. Cartwheel-like configurated radial striaies as well as fine cystoid changes in macula were seen in fluorescein angiography (FA) of the right eye and diffuse punctate filling defects were revealed in FA of the left eye [Fig. 2]. Bilateral foveal retinoschisis was evident on optical coherence tomography (OCT) [Fig. 3]. Bilateral subnormal rod responses in scotopic electroretinogram (ERG) and subnormal flicker responses were present; however, they were more pronounced in the left eye. Bilateral negative maximum responses in scotopic ERG were also detected. Besides, a selective decrease in b-wave amplitude on photopic cone responses which is characteristic for retinoschisis was evident for the subject.

In order to avoid the potential risk of retinal detachment, argon laser photocoagulation was performed around the peripheral retinoschisis lesion in the left eye [Fig. 1]. After the second year of follow-up visual acuity remained stable, also FA [Fig. 3] and OCT [Fig. 4] revealed no progression in time.

Discussion

The constant diagnostic feature of XLRS is bilateral foveal retinoschisis, whereas peripheral retinoschisis may be present approximately half of the affected male patients. Tangential splitting of the neurosensory retina caused by the defect of cellular adhesion and cell–cell interactions within the inner nuclear layer is main histopathological appereance. However, disease is commonly presented with reading difficulties due to the foveoschisis in school-age children, it can be diagnosed with nystagmus or strabismus in infancy. Prognosis is poor because of the progressive maculopathy, moreover sudden visual loss due to retinal detachment or intravitreal hemorrhage may be seen in cases with peripheral retinoschisis.1,3

Clinical photographs of retina and cross-sectional images of OCT help confirming the diagnosis and determining the management of the disease. Existence of mild window defects and absence of fluorescein leakage on FA as well as presence of a selective decrease in b-wave amplitude on both scotopic and photopic testing of ERG are also characteristic findings for differential diagnosis.1,2,5,6 Confirming RS1 gene mutations on genetic molecular testing is an emerging diagnostic parameter for the affected patients without a family history.3,10

However most of the retinoschisis cavities remain stable and rarely extend to the posterior pole, many authors suggest laser prophylaxis. Malagola et al.7 reported the success of prophylactic argon laser photocoagulation in the asymptomatic stage of bullous retinoschisis with outer layer breaks. Gopal et al.4 also reported the beneficial effect of prophylactic photocoagulation in collapsing the schisis cavities. Successful treatment with laser photocoagulation in one of three cases of posteriorly situated retinoschisis-retinal detachment was reported by Ambler et al.8 Jo et al.5 published the data of the first case of spontaneous regression of retinoschisis as observed by OCT in the literature. On the other hand, some authors do not recommend prophylactic treatment of retinoschisis in the absence of combined retinal detachment; moreover, they

Department of Ophthalmology, Dokuz Eylul University School of Medicine, and 1Dr. Behet Uz Children’s Hospital, Izmir, Turkey

Correspondence to: Prof. Nilufer Kocak, Department of Ophthalmology, Dokuz Eylul University School of Medicine, Inciralti, Balcova 35340, Izmir, Turkey. E-mail: nkocak@yahoo.com

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stated the increased risk of retinal tears and iatrogenic retinal detachments after the heavy photocoagulation burns.\textsuperscript{[4,6,9]} In the present case, we performed prophylactic argon laser photocoagulation to the posterior margin of retinoschisis cavity. At the end of the second year of follow-up, both fundoscopic examination and visual assessment remained stable. Although there is no proven guideline for both the laser prophylaxis and the treatment of retinoschisis, laser photocoagulation, cryotherapy, scleral buckle, and parsplana vitrectomy are considered as the main treatment options especially in cases with vitreous hemorrhage or retinal detachment.

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