Spectral Domain Optical Coherence Tomography Findings in Two Cases of X-Linked Juvenil Retinoschisis

X'e Bağlı Juvenil Retinoskizisli İki Olgunun Spektral Domain Optik Koherens Tomografi Bulguları

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ABSTRACT X-linked juvenile retinoschisis (XLRS) is a progressive bilateral disease that is probably present at birth and has been manifested by loss of vision in the early childhood. Due to X-linked transition the disease is seen predominantly in young boys. In this study we report spectral domain optical coherence tomography (SD-OCT) findings of XLRS in two cases of ten years old boy. One patient demonstrates typical spoke-wheel patern and the second patient demonstrates cystic macular appearance on fundus examinations in both eyes. Although there is different ophthalmoscopic feature between two cases, SD-OCT shows the retinoschisis cavities directly and makes the diagnose of the disase easy with high resolution imaging.

Key Words: Tomography, optical coherence; retinoschisis

ÖZET X'e bağlı juvenil retinoskizis (XLRS), muhtemelen doğumda var olan, erken çocukluk döneminde görme azlığı ile kendini gösteren, bilateral, ilerleyici bir hastalıktır. X'e bağlı geçiş şekli nedeniyle hastalık esas olarak genç erkeklerde görülür. Bu çalışmada XLRS'li 10 yaşında iki erkek olgunun spektral domain optik koherens tomografi (SD-OKT) bulgularını sunuyoruz. Fundus incelemesine göre her iki gözde, bir hastada tipik bisiklet tekeri görünümü, diğer hastada kistik makula görünümü mevcuttu. İki olgu arasında oftalmoskopik görüntüde farklılık olmasına rağmen, SD-OKT retinoskizis boşluklarını belirgin olarak göstermekte ve yüksek çözünürlüklü görüntüleme sayesinde hastalığın tanısını kolaylaştırmaktadır.

Anahtar Kelimeler: Tomografi, optik koherens; retinoskizis

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X-linked juvenile retinoschisis (XLRS) was first described in 1898 in two affected brothers by the Austrian ophthalmologist Josef Haas, has recently become better understood as a mutation of the XLRS1 gene on the short arm of the X chromosome (Xp22).[1,2] Characteristic features include mild to severe loss in central vision, radial streaks arising from foveal schisis and splitting of inner retinal layers. Splitting in the nerve fiber layer (NFL) attributed to Müller cell dysfunction was noted on histological studies and was observed in vivo with OCT by Eriksson et al.[3,4] In this study, we used SD-OCT (RS 3000, Nidek, Japan) to examine the foveal areas in two patients with juvenile retinoschisis.

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CASE REPORT

A 10 year old boy (Case 1) was presented for evaluation of bilaterally reduced visual acuity without acute visual loss. The patient's father reported that the boy had experienced poor vision for a few years. His best corrected visual acuity (BCVA) was 20/100 in both eyes with no improvement on manifest refraction or pinhole testing. The anterior segments were unremarkable. Fundus examination showed typical spoke-wheel pattern at the central macula in both eyes (Figure 1). Peripheral retinoschisis was present at the lower temporal quadrant in the left eye. There was no leakage at the macula in the early and late phase of fundus fluorescein angiography (FFA). The SD-OCT exams demonstrated findings consistent with XLRS (Figure 2).

Another 10 year old boy (Case 2) was presented for evaluation with the same complaint. His BCVA was 20/50 OD and 20/100 OS with no improvement on manifest refraction or pinhole testing. The anterior segments were also unremarkable. Fundus examination showed cystoid maculopathy at the fovea in both eyes (Figure 3).

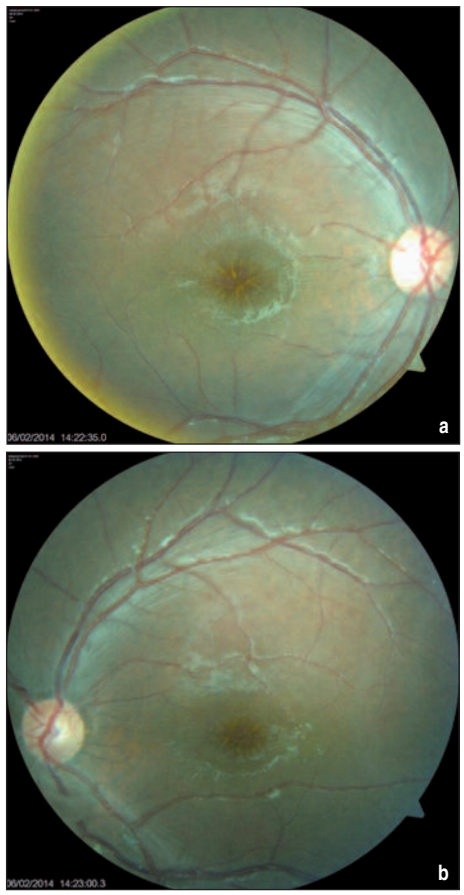

FIGURE 1: Fundus photographs of Case 1. The right **a)** and the left **b)** eye demonstrates a spoke-wheel appearance.

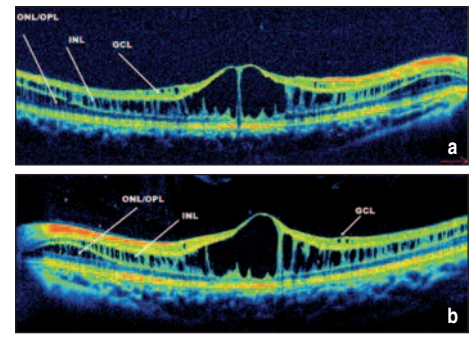

FIGURE 2: SD-OCT images of Case 1 taken through the center of the fovea. Both eyes demonstrate a large central cyst that extends from the nerve fiber layer to the outer retina. The retina is split into 3 layers by schisis cavities at the inner nuclear layer (INL) and outer nuclear layer (ONL)/outer plexiform layer (OPL), with retinal tissue remnants connecting each of the layers. There are small cysts at the ganglion cell layer (GCL). The schisis involves the full extent of the parafoveal region. **a)** Right eye and **b)** Left eye.

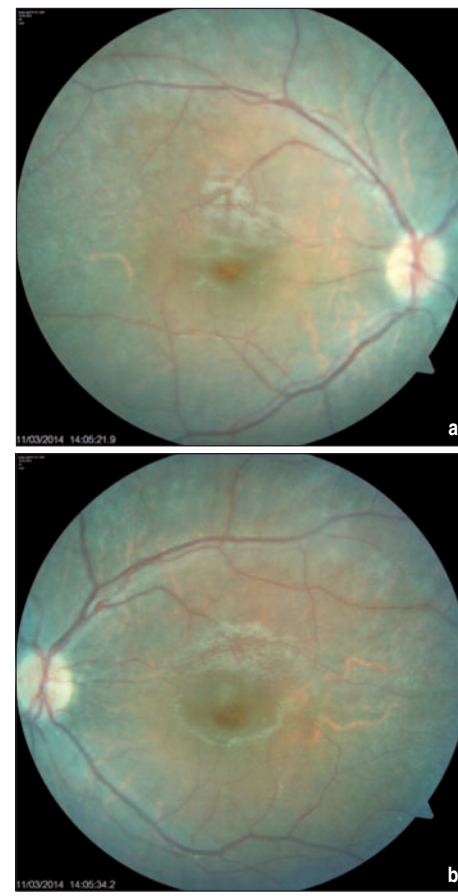

FIGURE 3: Fundus photographs of Case 2. The right eye **a)** and the left eye **b)** demonstrates cystic appearance.

There was no leakage on FFA. The SD-OCT exams demonstrated findings consistent with XLRS (Figure 4).

DISCUSSION

The diagnosis of XLRS typically is dependent on a careful fundus examination. The hallmark of the disease is the presence of a spoke-wheel pattern in the macula on high magnification ophthalmoscopy in patients younger than 30 years of age.[1] The spoke-wheel pattern varies in severity and might be difficult to detect or exclude especially in young boys with limited cooperation even by experienced clinicians, but OCT is helpful for confirming the presence of the schisis cavity directly.[5]

OCT has changed the diagnostic approach for XLRS and nowadays SD-OCT is the major diagnostic technique for this disease. SD-OCT has several advantages over time-domain OCT. Although the axial resolution of the two technologies is comparable, the advantage of SD-OCT is in the speed of image acquisition. SD-OCT imaging allows to distinguish the ganglion cell layer (GCL), the inner plexiform layer, the inner nuclear layer (INL), the outer nuclear layer (ONL), outer plexiform layer (OPL), the external limiting membrane, the photoreceptor inner segments, the outer segments, the retinal pigment epithelium, and in pathologic cases, Bruch's membrane.[6]

Several publications describing the OCT characteristics of XLRS have noted different findings for this condition. Azzolini et al. reported OCT findings in 3 cases of XLRS showing a macular cleavage plane in the outer retinal layers as well as in the NFL.[7] Gao et al. reported OCT findings in two cases of XLRS showing that the foveal cystoid separation is not located in the NFL, but is in the outer plexiform layer.[8] These reports were using time-domain OCT, so there were some difficulties

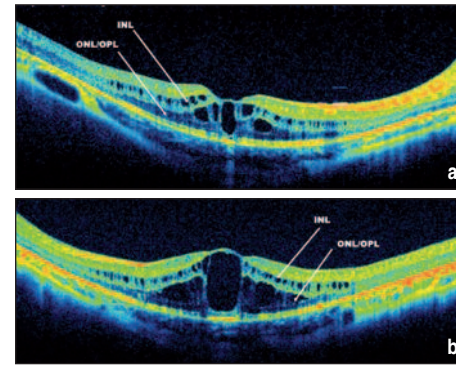

FIGURE 4: SD-OCT images of Case 2. The left eye **b)** demonstrates a large cyst at the fovea and two schisis cavities at the INL and ONL/OPL that is confined to the fovea. The right eye **a)** demonstrates multiple cystic spaces at the fovea. Two schisis cavities, at the INL and ONL/OPL, also are present, but are confined to the fovea and are less prominent than that of the fellow eye.

in detecting which retinal layer is split. In recent years SD-OCT resolved the long-standing histologic debate about which retinal layer is splitting. Leng reported two cases of XLRS with different SD-OCT finding in which one case the retina was split into 3 layers and in the other case there were multiple small cystic spaces in multiple retinal layers without disruption of the overall structure of the retina.[9] In our Case 1, the retina is split into 3 layers by schisis cavities at the INL and ONL/OPL and the schisis involves the full extent of the parafoveal region in both eyes. In Case 2, there are also two schisis cavities, at the INL and ONL/OPL, but are confined to the fovea. As in our cases Yu et al. reported a large series of XLRS and showed the foveomacular schisis seems to occur predominantly at the INL, occasionally at the ONL/OPL, and only rarely at the RNFL.[10]

In conclusion, the schisis cavity can ocur in a number of different layers of neurosensory retina in patients with XLRS. SD-OCT allows enhanced visualization of retinal layers with high resolution imaging and is important for the diagnose and follow-up examinations of the disaese.

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