

Multimodal imaging in retinoschisis X-linked.

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Abstract

To report retinal alterations of a X-Linked Juvenile Retinoschisis (XLRS) case of with multimodal imaging. This case report highlights the role of multimodal imaging examining vascular and structural alterations under this condition through non-invasive imaging techniques to help with making the diagnosis and follow up.

Keywords: Diagnosis, multimodal imaging, vitro retinal disorder, optical coherence tomography

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Introduction

Congenital Retinoschisis (XLRS) is a rare bilateral vitreoretinal disorder transmitted as an X linked recessive trait associated with a mutation of the *XLRS1* gene located on the short arm of the X chromosome, Xp22 [1]. The estimated prevalence is up to 1 in 20,000 with a bilateral appearance in 40% of the patients [2]. Clinically, the patients show typically a cystic-like stellate maculopathy or a foveal schisis and peripheral retinoschisis of various degrees of severity on fundoscopy [3]. Other authors described this condition in Optical Coherence Tomography (OCT) and OCT angiography (OCTA) as splitting of inner retinal layers and perifoveal microvascular changes, more evident in the deep vascular plexus [4]. The splitting of the inner retinal layers, mainly involving Inner Nuclear Layer (INL) followed by Outer Plexiform Layer (OPL) and ganglion cell layer (GCL), determines the formation of foveal schisis that leads to early bilateral visual loss [5]. In addition, XLRS may be complicated

by retinal detachment, vitreous or intraretinal hemorrhage and vascularized vitreous veils [6,7]. The purpose of our study is to describe XLRS patient with multimodal imaging modality including Spectral-Domain OCT (SD-OCT), En-face OCT, OCTA and fundus autofluorescence.

Case Presentation

We will take into consideration and describe the case of a 19 year-old male with juvenile retinoschisis whose Best Correct Visual Acuity was 20/50 in the right eye and 20/40 in the left one. Multicolor images (Heidelberg Engineering, Heidelberg, Germany) detect cartwheel-like macular degeneration (Figure 1A). Auto fluorescence (Heidelberg Engineering, Heidelberg, Germany) images reveal cystoid hyper auto fluorescence spaces (Figure 1B). Spectral Domain OCT (heidelberg spectral domain Optical Coherence Tomography (OCT) shows cystoid spaces in Ganglion Cell Layer (GCL), Inner Nuclear Layer (INL), Outer

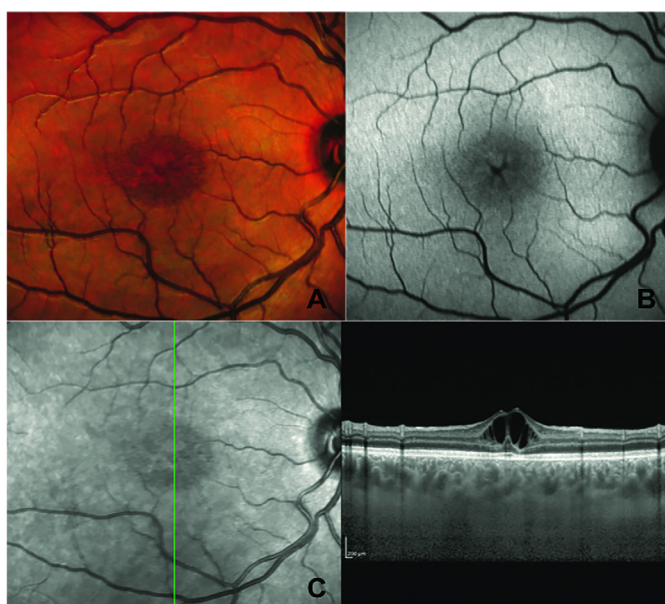


Figure 1: a) Multicolor images of the right eye demonstrate cartwheel-like macular Degeneration. b) Autofluorescence images reveal cystoid hyperautofluorescence spaces c) Spectral Domain Optical Coherence Tomography shows cystoid spaces in the inner layers

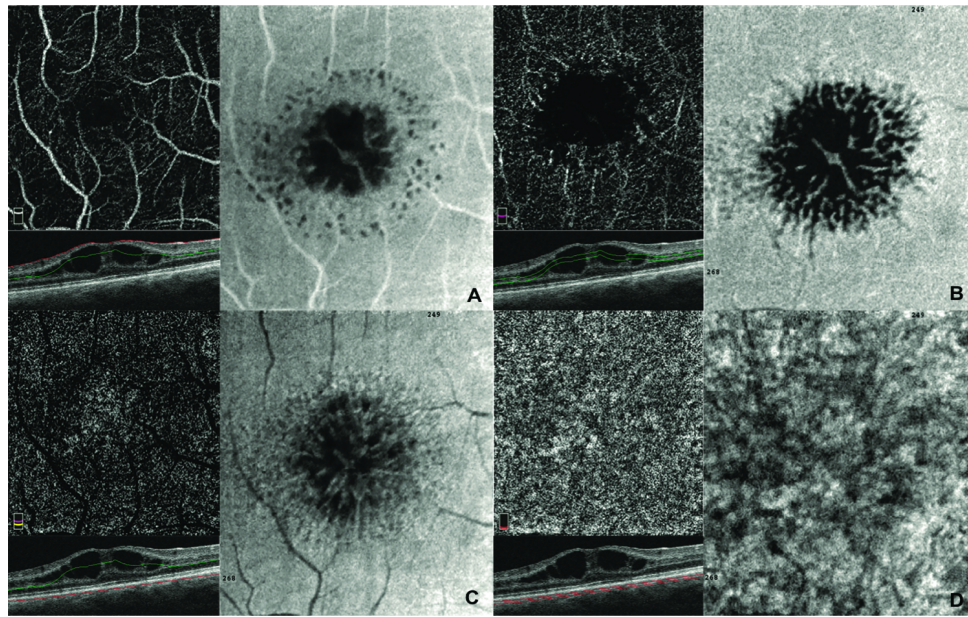


Figure 2: (a,b,c,d) OCT angiography and En face OCT of right eye show an increase of foveal A vascular zone in both superficial and deep vascular plexus, coral shape image in the inner retina and hypo hyper-reflectivity of the choroid layer.

Plexiform Layer (OPL) and the Outer Nuclear Layer (ONL). In addition, central macular and choroidal thickness have increased (Figure 1C). In agreement with other authors, OCTA (Angiovue System, Optovue Inc. Fremont, CA, USA) shows an increase of the foveal avascular zone in both superficial and deep vascular plexus and the presence of teleangiectasias (Figure 2A and 2B) [4-8]. In Face images reveal the presence of reflective areas that appear like a petaloid or coral shape without coriocalpilaris alteration (Figure 2A- 2C). Choriocalpilaris images demonstrate hypo hyper-reflective areas due to a shadow effect without the presence of any vascular alteration (Figure 2D).

Discussion

Multimodal imaging enables to provide different but complementary information on X linked juvenile retinoschisis affecting both the inner and outer retinal structures. In our study, the SD-OCT analysis shows that schisis involves the GCL, INL, OPL and ONL. Other reports have highlighted that the most prevalent area of schisis is INL followed by GCL and OPL [5-9] Nan-Kai Wang et al. have noted the typical auto fluorescence pattern of hyper- and hypo-auto fluorescence in the macular area [10] Our findings show the appearance of macular schisis visible to auto fluorescence like a cystoid hyper auto fluorescence spaces, expression of changed light transmission. In agreement with previous studies, we have observed on OCTA images an increase of foveal avascular zone in both superficial and deep neurovascular plexus and we have highlighted vascular abnormalities such as teleangiectasias like micro-vascular changes and capillary dropout rarefaction. These alterations of perifoveal microvasculature are described especially in the perifoveal deep vascular plexus due to the presence of schisis, accounting for possible biomarkers of disease prognosis. Moreover, Stanga et al. described a foveal petaloid a reflective area [8] and Tomoyo Yoshida-Uemura et al. described characteristic schisis patterns hyporeflexive holes and a spoke-like pattern respectively in parafoveal and foveal region

in GCL, a reticular pattern in INL and hyporeflexive polygonal cavities in OPL [9-11]. In our case, foveal schisis appears on en-face OCT like a petaloid or coral shape without coriocalpilaris alteration. In the present case, multimodal imaging will shed light on the *in vivo* structure of this rare condition offering more tools for its diagnosis, identifying typical features and helping in the follow up. Further experience with OCTA may clarify the precise role of this technology in diagnosing and managing retinal diseases.

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