X- linked Juvenile Retinoschisis: A Case Report

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PURPOSE

Describe a case of x-linked Juvenile Retinoschisis(XLRS)

INTRODUCTION

XLRS occurs exclusively in males and is characterized by visual loss that begins in early childhood. Is a retinal dystrophy due to pathogenic variants in the RS1 gene.It is characterized by separation of the retinal layers and the hallmark of the disease is a foveal spoke-wheel appearance.

METHODS

Medical records review

RESULTS

A 17 year old boy with vision loss since childhood. Ophthalmologic examination, he had best-corrected visual acuity of 20/60 in both eyes. In the fundus examination of both eyes he presented fovea in a spoke-wheel.
Optical coherence tomography (OCT) the presence of foveoschisis, parafoveal schisis, and foveal atrophy was evaluated, defects in the microstructures of the outer retinal photoreceptors were also observed. Fundus autofluorescence findings were spoke wheel pattern, foveal hyperautofluorescence and central signal reduction.

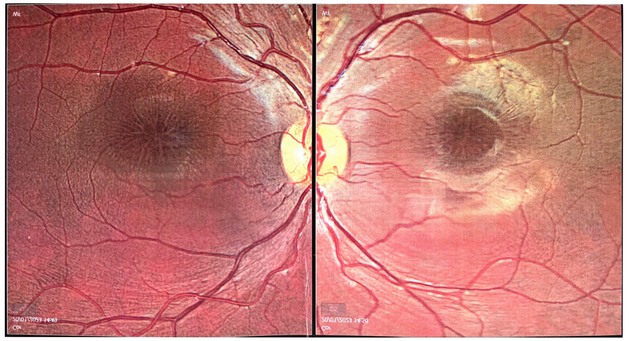

Fig. 1 and 2: retinography of the both eyes

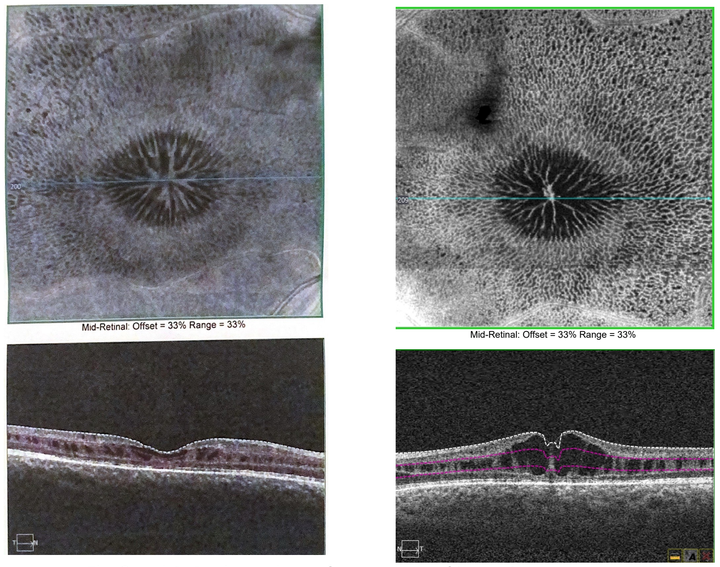

Fig. 3 and 4: Angiography en face analysis of the both eyes

DISCUSSION

XLRS is a hereditary and recessive degeneration, mutations in the XLRS1 gene, responsible for encoding retinoschisin. Defects or absence of its secretion can reduce adherence between the nerve fiber layer and the rest of the sensory retina, forming cystic cavities.
XLRS manifests itself with a decrease in visual acuity between five and ten years of age, and evolves with progressive visual loss during of life.
OCT scans show changes in the evolving foveolar architecture, resulting from cysts in the nerve fiber layer. These cystic formations are also found in the retinal periphery.
The electroretinogram is important in the diagnosis of this disease. The amplitude of wave - B its greatly reduced.
Treatment aims to contain damage, yet the disease is an attractive target for gene therapy due to its monogenic nature and promising preclinical studies.

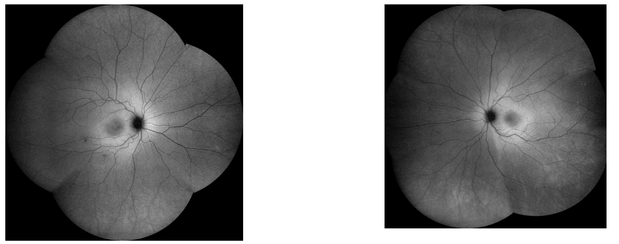

Fig. 5 and 6: Fundus autofluorescence of the both eyes

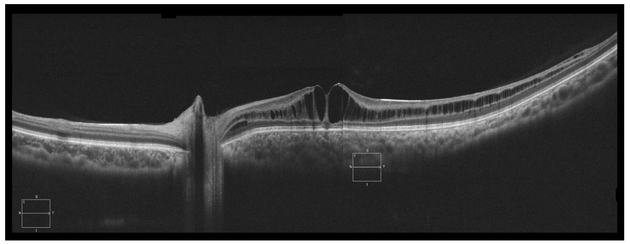

Fig. 7 OCT - Junction of 3 cuts of 9 mm HD line of the left eye