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X-Linked RetinoschisisCase Report

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Abstract
X-linked Retinoschisis (XLRS) is one of the most common macular degenerations in young male. It is characterized by a high degree of clinical variability.The semiology of the examination of the fundus, the OCT and the electroretinogramallowtodiagnose this maculopathy.
DNA analysis confirms the diagnosis of macular retinoschisis linked to X and provides the opportunity for genetic counselling for the patient and family.
Keyword:*Juvenile retinoschisis, X-linked retinoschisis.*

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I. Introduction

X-linked Retinoschisis (XLRS) is one of the most common macular degenerations in young male. This disease is characterized by a high degree of clinical variability [1], [2].

The Clinical signs of this disorder may include a macular star with or without peripheral retinoschisis. In addition, Separation occurs in the retina, mainly at the nerve fiber layer. It is responsible for a decrease in the visual acuity of varying significance and is slowly progressive, generally appearing during the first decade. Although there is no effective treatment to stop the progression of the maculopathy, clinical management focuses on treating the amblyopia and the surgical correction of some complications[3].

II. Observation

An 10-year-old boy, with no pathological history, consults for a decreaseprogressive visual acuity. At the eye examination, his best corrected visual acuity was 3/10 in both eyes on the Snellen chart. A slit-lamp examination showed a normal anterior segment in each eye, normal intraocular pressure, and a clear lens. Gonioscopy revealed open angles in both the eyes without abnormalities. The eye fundus showed typical fovealschisis in a stellate pattern to both eyes associated with a peripheral schisis in the extreme periphery to the right, and in the middle periphery to the left.

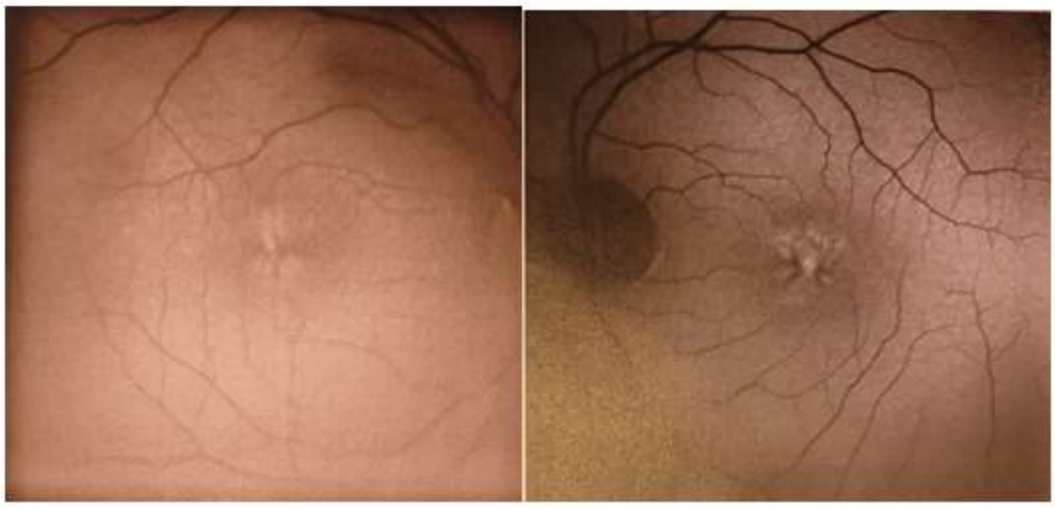

Figure 1: Fundus autofluorescence (FAF) of the right and left eye showing the fovealschisis. with radiating spokes of retinal splitting

The OCT showedintraretinal cysts in the external and internal nuclear layers, as well as some cysts in the ganglion cell layer. These cysts are clearly visible at the macula by autofluorescence. In addition, a treatment with AZOPT, at a frequence of one drop 3 times a day, has been initiated as it appears that carbon dioxide inhibitors may have some effect on cyst reduction.

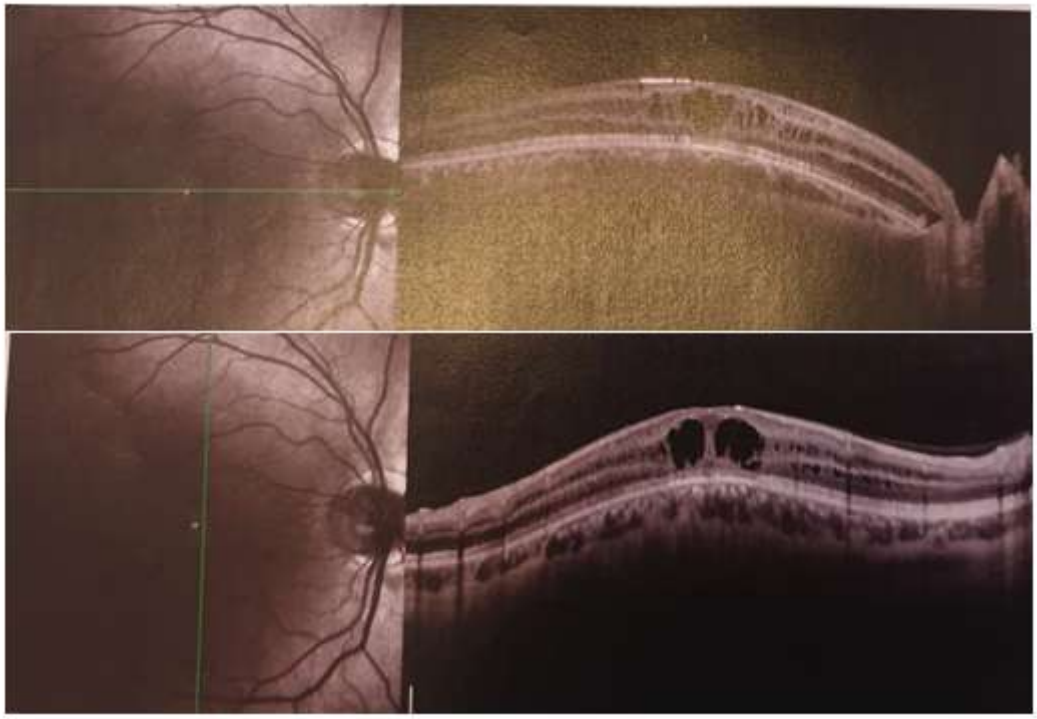

Figure 2: Spectral-domain optical coherence tomography of the RIGHT: Horizontal and vertical section demonstrates fovealschisis withe increased thickness in the fovea.

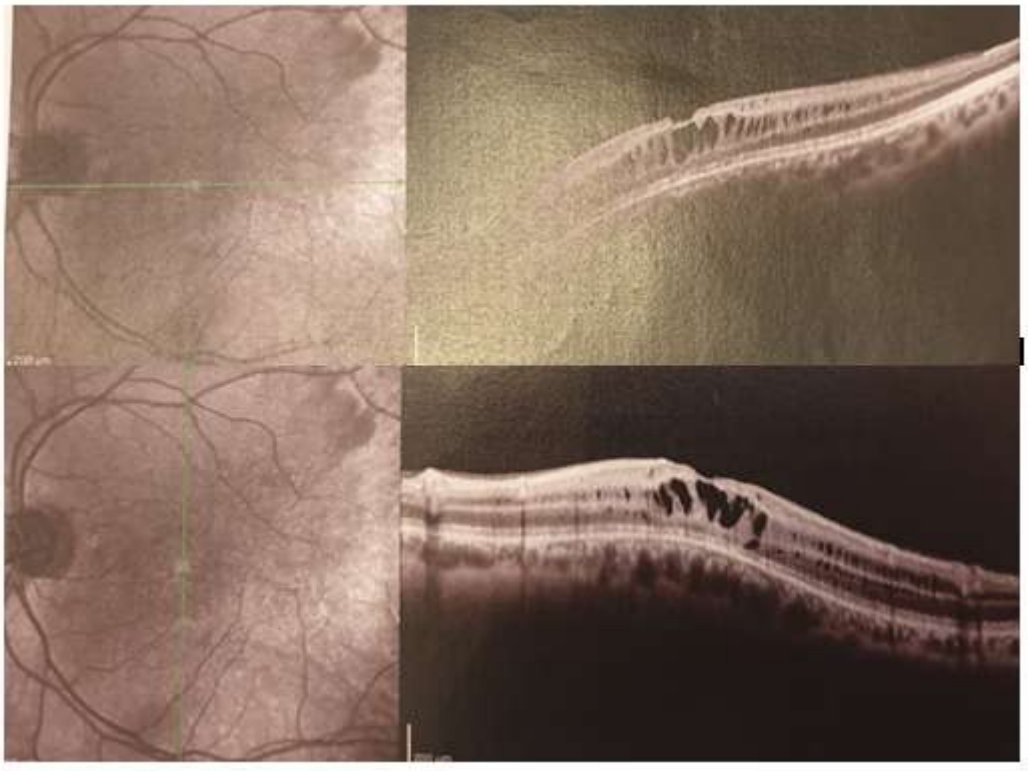

Figure 3: Spectral-domain optical coherence tomography of the LEFT : Horizontal and vertical section demonstrates fovealschisis withe increased thickness in the fovea

A electrophysiological assessment showed a major dysfunction, mainly b-wave reduction on the full-field electroretinogram and damage to the conduction of the visual evoked potentials. Color vision revealed dyschromatopsia without an axis.

His family tree tells that he's in the 2th position of a sibling of 3 children including 2 boys. His older brother would not have developed the disease. His parents have no apparent blood connection.

We discussed with parents the mode of transmission of retinoschisis associated with X.

III. Discussion

Juvenile retinoschisis linked to X is a congenital maculopathy secondary to an abnormal retinal cleavage. This is the most common cause of juvenile macular degeneration in boys. Transmission is recessive and linked to the X chromosome, penetration is complete and expressiveness is variable.

There is a great clinical heterogeneity from one individual to another, but also within the same family. The diagnosis is most often made in school-aged boys before the appearance of difficulties while reading. Juvenile retinoschisis linked to X is characterized by a bilateral stellar microcystic macular reshaping, centered on the fovea, with a 'wheel spokes' appearance, which is found in 98% of cases [4], associated in 40-50% of cases with peripheral retinoschisis, most often located in the lower temporal [5]. This superficial bubble has a spontaneous tendency to reapplication.

It is not a retinal detachment and does not require surgery or laser photocoagulation, but regular monitoring. In 40% of cases, juvenile retinoschisis linked to X is complicated by intravitreal or intracystic hemorrhage by rupture of a vessel in bridge or by a secondary neovascularization. The major complication, however, is retinal detachment, which is estimated to occur between 5 and 22% of the cases[6].

It can be either a rhegmatogenic detachment or a tractional detachment, and is more frequently seen in children under 10 years of age. Fluorescein angiography is not very contributory in the macular retinoschisis linked to X. Angiography is most often normal, contrasting with the characteristic aspect when examining the fundus [7].

The Optical coherence tomography (OCT) confirms the diagnosis of macular retinoschisis linked to X, it shows schisis in the superficial neural retina and thinning of the retina. In most school-aged children, there are often small cystic-like spaces peri-foveal and large cystic-like spaces within the fovea. After adolescence, the cystic spaces may not be as evident because of flattening of cysts with increased age.These cystic spaces occur predominantly in the nerve fiber layer. OCT can reveal areas of schisis that may not be visible on fundus examination[8].

Electroretinogram can also contribute to diagnosis by showing decreased b-wave in scotopic and photopic[9]. Other studies, such as indocyanin green angiography, visual field examination, or colour vision testing, are not very helpful for the positive diagnosis of X-related macular retinoschisis.

Currently, there is no specific treatment for retinoschisis. The use of dorzolamide topically with improved OCT and visual acuity has been published for small series of patients, but further studies are needed to validate these results [11]. Laser photocoagulation and vitreoretinal surgery may be indicated in case of complications.

Genetically, the mode of transmission is recessive, linked to the X chromosome, and this condition rarely affects women. Rarer forms of autosomal dominant or autosomal recessive transmission have been described [10-11-12].

The gene involved in the classic X-linked form codes for retinoschisin, a 224 amino acid protein present in the photoreceptor layer of the inner retinal layers. Retinoschisin contributes to the architecture of the cell layers of the retina, by participating in its synaptic organization and is believed to be associated with the Na+/K+-ATPases of the photoreceptor membrane and bipolar cells [13-14]. The research work that followed the discovery of the gene involved in 1997 (15) paved the way for gene therapy trials in mouse models with an invalidated RS1 gene [16-17].

IV. Conclusion

X-linked retinoschisis may exhibit significant clinical variability and must be reported in case of macular alterations in a male child but also in adults.

The diagnosis of the least severe forms is not obvious, and the delay between the first symptoms and the diagnosis is often considerable. DNA analysis confirms the diagnosis and provides the opportunity forgenetic counselling to the patient and family.

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