

## Complicated Juvenile Sex-Linked Retinoschisis

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### Abstract

### Case Report

**Introduction:** Juvenile sex-linked retinoschisis is the most frequent of juvenile macular degenerations. It affects the male sex with variable expressiveness. The first clinical manifestations generally appear during the first decade, characterized by visual disturbances with a decrease in visual acuity of variable importance. Its evolution can be complicated by cataracts, strabismus, vitreous haemorrhage and retinal detachment. **Case Report:** A 14-year-old adolescent presented with severe bilateral low vision with nystagmus from early childhood, referred for etiological diagnosis. The examination of the right eye showed a visual acuity at negative light perception with a white cataract obstructing the eye fundus access. The examination of the left eye found visual acuity at hand motion, a posterior subcapsular cataract and at the eye fundus: a diffuse chorioretinal atrophy with a stellar macular changes. Ocular ultrasound of the right eye finds a retinal detachment on suspected retinoschisis without intravitreal haemorrhage, and a probable juvenile retinoschisis in the left eye. Macular OCT find in the left eye a cleavage in the retinal thickness evoking severe retinoschisis. The electrophysiological tests: electroretinogram, electro-oculogram and visual evoked potentials; were very altered in both eyes. **Conclusion:** Juvenile sex-linked retinoschisis is a serious condition that affects young boys, responsible for low vision due to macular damage. Through this rare case, we emphasize the interest of early diagnosis of juvenile retinoschisis and regular retinal monitoring in order to watch for progression to low vision or even blindness.

**Keywords:** juvenile sex-linked retinoschisis; low vision; retinal detachment; complications; management.

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## INTRODUCTION

Juvenile sex-linked retinoschisis is the most common type of juvenile macular degeneration [1]. It affects the male sex with variable expressiveness. The first clinical manifestations generally appear during the first decade. It is manifested by visual disturbances with a decrease in visual acuity of variable importance. A nystagmus can be observed in severe cases [2]. Its evolution can be complicated by cataracts, strabismus, vitreous haemorrhage and retinal detachment [3]. In severe cases, schisis affects the entire thickness of the retina, leading to low vision or even blindness. We report the case of a young adolescent presented with severe bilateral low vision with nystagmus from early childhood, referred for etiological diagnosis, in whom a diagnosis of juvenile sex-linked retinoschisis complicated by retinal detachment and total cataract was retained. Through this rare case, we highlight the interest of early diagnosis of juvenile retinoschisis and

regular retinal monitoring in order to watch for progression to low vision or even blindness.

## CASE REPORT

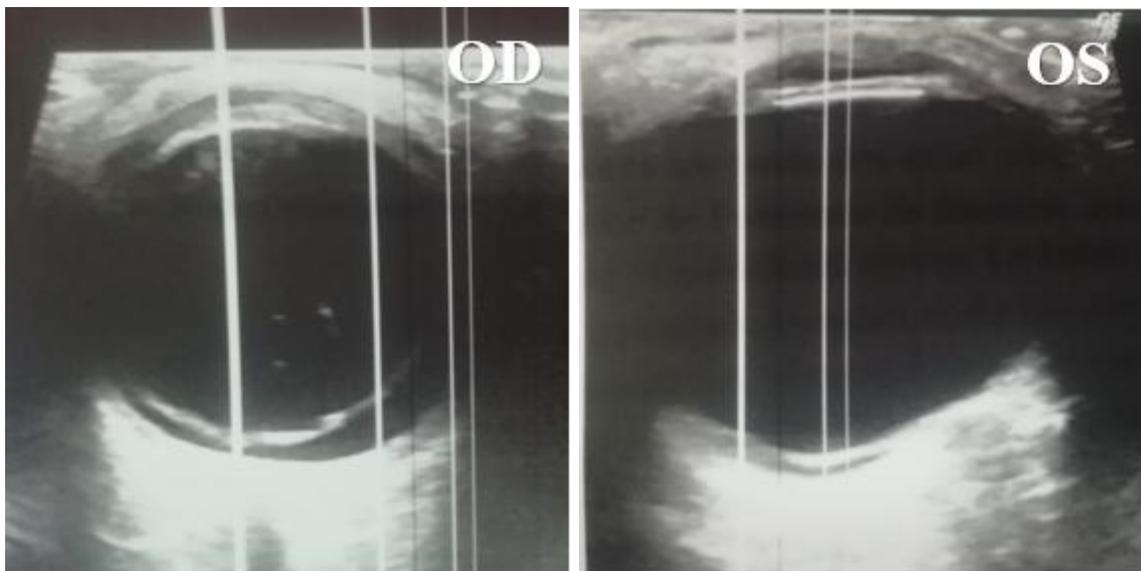
It is a 14-year-old adolescent, who has complained of low vision with nystagmus for over 8 years, for which he was referred for etiological diagnosis. The inspection finds a permanent horizontal binocular nystagmus with no fixation and a permanent head torticollis. His visual acuity was at NLP in OD and hand motion in OS, unimproved. The slit lamp examination of the right eye found a clear cornea with good anterior chamber, normal intraocular pressure and a total white cataract blocking the eye fundus examination. The examination of the left eye found a posterior subcapsular cataract with a clear vitreous, and in the eye fundus (Figure-1) a diffuse chorioretinal atrophy, an absent foveal reflex with aspect stellar macular changes. The examination of OS retinal periphery was difficult hampered by nystagmus.



**Fig-1: Color fundus photography of the left eye showing diffuse chorioretinal atrophy and absent foveal reflex with aspect of stellar macular degeneration**

Ocular ultrasound (Figure-2) showed in the right eye, a thick hyperechoic membrane with limited mobility evoking an old retinal detachment without intravitreal haemorrhage or visible tears. In the left eye, ultrasound showed a small localized and hyperechoic

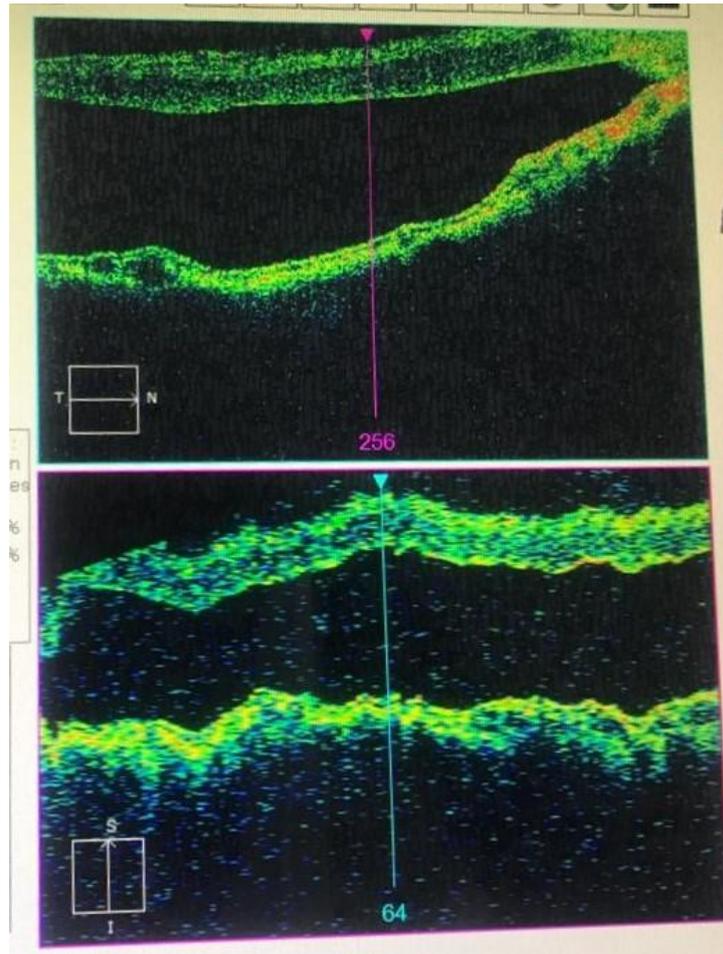
membrane with a thinner thickness suggesting a juvenile retinoschisis. Retinal angiography was difficult to perform due to the permanent involuntary movements of both eyes.



**Fig-2: Ocular ultrasound showing in OD: a thick and hyperreflective membrane evoking an old retinal detachment without intravitreal haemorrhage or visible tears, in OS: a small localized hyperechoic membrane with thinner thickness evoking a probable juvenile retinoschisis**

The macular OCT (Figure-3) performed in OG, of imperfect quality due to absence of fixation, showed an evident cleavage in the retinal thickness evoking a severe retinoschisis. The electrophysiological tests: electroretinogram, electro-oculogram and evoked potentials and very, were altered in both eyes. At the therapeutic level, and despite the profound visual impairment and the low hope of functional recovery, a cataract surgery of the left eye was indicated to allow

regular retinal monitoring, in order to watch for a possible secondary retinal detachment on juvenile retinoschisis. In the right eye, no treatment has been decided given the long delay of the retinal detachment becoming rigid, the profound alteration of electrophysiological parameters and the negative visual acuity. Orthoptic amblyopia treatment of the left eye has also been indicated.



**Fig-3: Macular OCT of the left eye (of imperfect quality due to the absence of fixation) showing a cleavage in the retinal thickness evoking a severe retinoschisis**

## DISCUSSION

Juvenile sex-linked retinoschisis is the most common type of juvenile macular degeneration. It is a rare hereditary dystrophy of X-linked recessive inheritance. It affects the male sex with variable expressiveness, the gene in question being XLR1, located in position Xp22.2-p22.1 [4]. It results from a cleavage within the layer of nerve fibers secondary to the degeneration of Müller's cells [5]. The diagnosis is often made at school-age when there is a visual acuity decrease. Macular retinoschisis is the pathognomonic sign [2]. Peripheral retinoschisis is encountered in 50% of cases in the inferior temporal region. The diagnosis of juvenile sex-linked retinoschisis can be made clinically, on the eye fundus aspect [6]. The scotopic electroretinogram shows a decrease in amplitude of the b wave while the negative a wave is relatively preserved; the photopic electroretinogram is normal [7]. OCT objective the schisis areas in the macular region. In addition, close interrogation most often reveals a family history consistent with X-linked transmission. Molecular analysis by direct sequencing of the RS1 gene makes it possible to detect mutations in approximately 90% of patients [4].

Juvenile sex-linked retinoschisis is a progressive pathology with severe prognosis. Patient follow-up must be close in order to detect and manage complications such as strabismus, cataracts, retinal detachment and vitreous haemorrhage [3]. The retinal detachment complicating juvenile retinoschisis is most often localized, it represents a rare complication. Surgical treatment can only be indicated in the progressive forms of retinal detachment [8]. In these cases, endo-ocular surgery is indicated allowing good reapplication of the retina, followed by laser photocoagulation of outer layer dehiscences [8, 9]. In some cases, anatomical success can be accompanied by an unfavorable evolution towards the eyeball phtysis, particularly in advanced forms with delayed management.

## CONCLUSION

Juvenile sex-linked retinoschisis is a serious condition affecting young boys, responsible for low vision due to macular damage. The clinical presentation is variable but the prognosis remains poor. The evolution can be marked by serious complications whose treatment is often difficult. Through this rare case, we emphasize the interest of early diagnosis of juvenile retinoschisis and regular retinal monitoring in

order to detect complications and watch for progression to low vision or even blindness.

**Conflicts of interest:** The authors declare no conflicts of interest.

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