

# X-Linked Juvenile Retinoschisis: A Case Report

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## Abstract

X-linked juvenile retinoschisis is a hereditary macular dystrophy that is transmitted in the X-linked recessive mode. Clinical signs include a macular star with or without peripheral retinoschisis responsible for decreased visual acuity. This study dealt with a 12-year-old boy who came in for a consultation for progressive decline in visual acuity. His distance visual acuity without correction was scored at 5/100; the right eye (RE) improved to 10/100 after the correction of a myopic astigmatism; and the unimproved left eye was scored at 20/100. The eye fundus showed perimacular radial lines without increased separation for the right eye with some microcysts and a macular hole on the left. An examination of the retinal periphery of both the eyes found inferotemporal retinal splitting. The electrophysiological assessment showed a major dysfunction on the electroretinogram.

**Keywords:** Degeneration, macula, retinoschisis, star

## INTRODUCTION

X-linked juvenile retinoschisis is one of the primary causes of macular degeneration in male children. It is characterized by a high degree of clinical variability.<sup>[1,2]</sup> Clinical signs include a macular star with or without peripheral retinoschisis. 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.<sup>[3]</sup>

## Case Report

This study was regarding a 12-year-old boy who came in for a consultation for progressive decline in visual acuity over 2 years. His distance visual acuity without correction was scored at 5/100; the right eye (RE) improved to 10/100 after the correction of a myopic astigmatism; and the unimproved left eye was scored at 20/100. 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 anomalies. The eye fundus showed perimacular radial lines without increased separation for the right eye with some microcysts and a macular hole on the left. An examination of the retinal periphery of both the eyes found inferotemporal retinal

splitting with a demarcation line corresponding to separation [Figure 1]. A vitreous examination did not show any significant change. Retinal angiography identified perimacular radial lines, with inferotemporal bilateral peripheral separation. Fluorescein injection did not show macular edema or vascular degeneration [Figure 2]. A electrophysiological assessment showed a major dysfunction, mainly b-wave reduction on the full-field electroretinogram [Figure 3] and damage to the conduction of the visual evoked potentials. Color vision revealed dyschromatopsia without an axis. An examination of six members of the family revealed a trisomy in the 5-year-old younger brother without the involvement of eye anomalies in parents and siblings. Molecular biological tests could not be performed.

## DISCUSSION

X-linked juvenile retinoschisis is the most common juvenile macular degeneration disease.<sup>[1,2]</sup> It only affects boys with variable expressivity. Its global prevalence varies between 1/5000 and 1/25,000.<sup>[4,5]</sup> The highest prevalence has been reported in Finland.<sup>[6,7]</sup>

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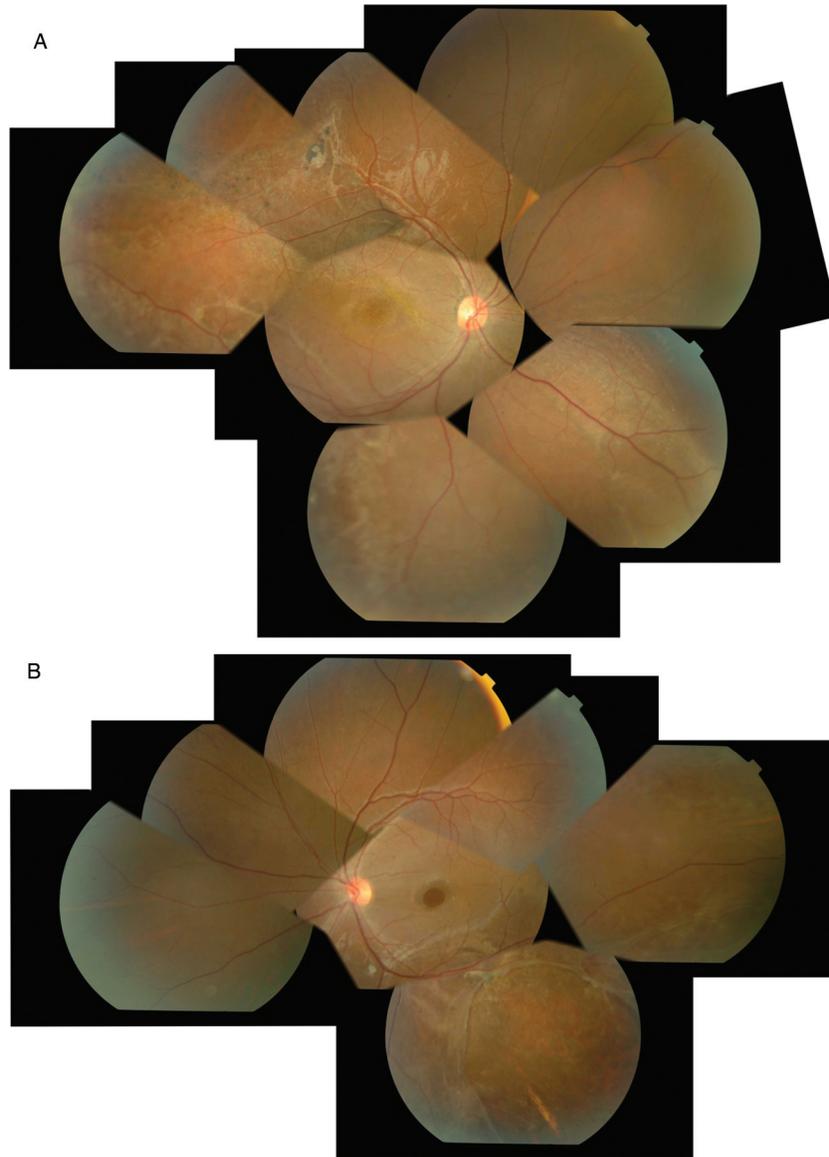


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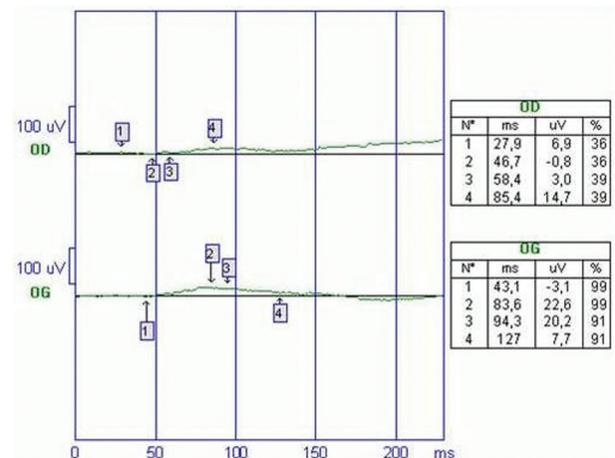
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**Figure 1:** (a) right eye (RE) fundus: perimacular radial lines, microcysts, and inferotemporal retinal splitting. (b) left eye (LE) fundus: macular hole and inferotemporal retinal splitting



**Figure 2:** Fundus fluorescein angiography (FFA) right eye (RE)



**Figure 3:** Electroretinogram: b-wave reduction

A decrease in visual acuity is common in most clinical presentations involving retinoschisis.<sup>[8-10]</sup> This decrease in visual acuity is often detected in boys between 5 and 10 years old due to reading difficulties in school. Visual acuity at this age is highly variable, but is generally above 20/100. In our case, the visual acuity in the best eye was 20/100 at 12 years old. Hypermetropia is a frequent finding but was not true in this case.<sup>[11]</sup>

A family examination is crucial. It makes it possible to either clarify the known family history or, conversely, to detect genetic abnormalities in the genealogy, as was the true for this case. Macular separation was the pathognomonic sign.<sup>[9,10,12]</sup> Sometimes, it may be lacking with the presence of divergent retinal radial folds from the foveola to the macular periphery; and in 50% of the patients, peripheral retinoschisis is inferotemporal, as was true for our case. Electrophysiological disturbances are related to neuroretinal damage. A possible involvement of foveolar Müller cells in the formation of the idiopathic macular hole has been suggested. This case could, thus, support the hypothesis of the participation of foveolar Müller cells in the pathogenesis of the macular hole.<sup>[12]</sup> The RS1 gene (MIM 300839) associated with the disease encodes retinoschisin, a 224 amino acid protein containing a discoidin domain as the major structural unit, an N-terminal cleavable signal sequence, and regions responsible for subunit oligomerization.<sup>[11]</sup>

## CONCLUSION

X-linked juvenile retinoschisis is a condition affecting young boys and is responsible for a visual handicap that is worsened by the occurrence of a macular hole.

## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

## Conflicts of interest

There are no conflicts of interest.

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