International Journal of Science and Research (IJSR)
ISSN: 2319-7064
Impact Factor 2024: 7.101

Bilateral X-Linked Juvenile Retinoschisis: A Case Report

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Abstract: ***X-linked juvenile retinoschisis (XLRS) is a rare hereditary retinal dystrophy that primarily affects young males, often leading to progressive visual impairment. We report a case of a 10-year-old boy presenting with a three-year history of bilateral, painless reduction in vision. A significant maternal family history suggested an X-linked inheritance pattern. While anterior segment examination was unremarkable, fundoscopic findings were subtle. Optical coherence tomography (OCT) played a pivotal role in the diagnosis, revealing characteristic intraretinal schisis cavities within the macular region of both eyes. The patient was managed conservatively with a focus on long-term monitoring for sight-threatening complications such as retinal detachment or vitreous hemorrhage. This case highlights the indispensable role of OCT as a non-invasive diagnostic "gold standard" in identifying XLRS when clinical signs are minimal, facilitating early intervention and familial genetic counselling.***

Keywords: X-linked juvenile retinoschisis, optical coherence tomography, macular schisis, pediatric ophthalmology, RS1 gene

1. Introduction

X-linked juvenile retinoschisis (XLRS) is a vitreoretinal dystrophy caused by mutations in the RS1 gene, which encodes the retinoschisin protein responsible for cell-to-cell adhesion within the retina[1]. Deficiency in this protein leads to the characteristic splitting (schisis) of retinal layers. XLRS typically manifests in the first decade of life with bilateral vision loss. Advances in imaging, particularly Optical Coherence Tomography (OCT), have revolutionized the diagnosis by allowing for the visualization of schisis cavities even in asymptomatic or clinically subtle cases[2].

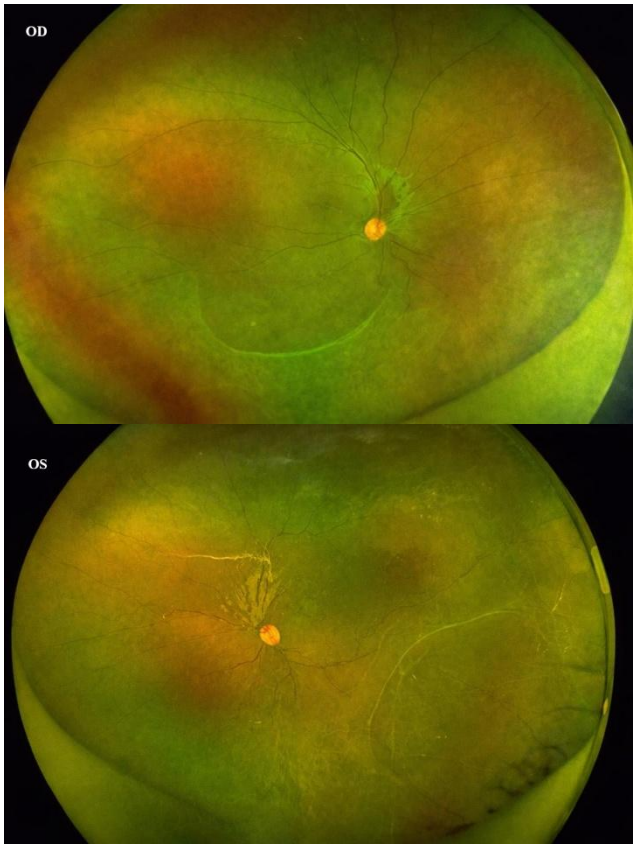
2. Literature Survey

The prevalence of XLRS is estimated between 1 in 5,000 to 1 in 25,000 males globally. While foveal schisis is present in nearly all cases, peripheral schisis occurs in approximately 50% of patients [3]. Disease is generally slowly progressive until young adulthood, it can be punctuated by sudden vision loss due to complications like vitreous hemorrhage [4]. Recent studies have also explored the use of carbonic anhydrase inhibitors to reduce cystic spaces, though conservative observation remains the primary management strategy for stable cases [5].

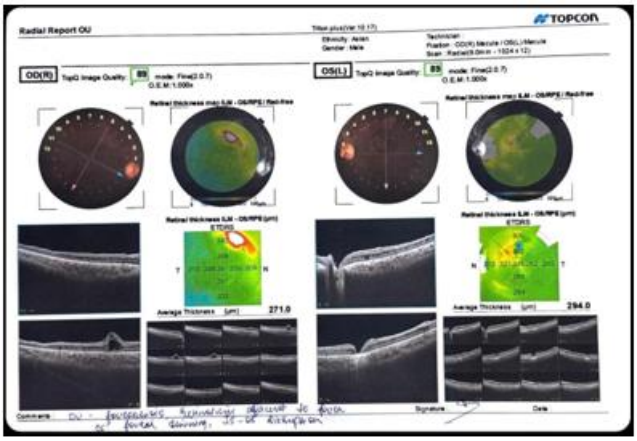
3. Case Report

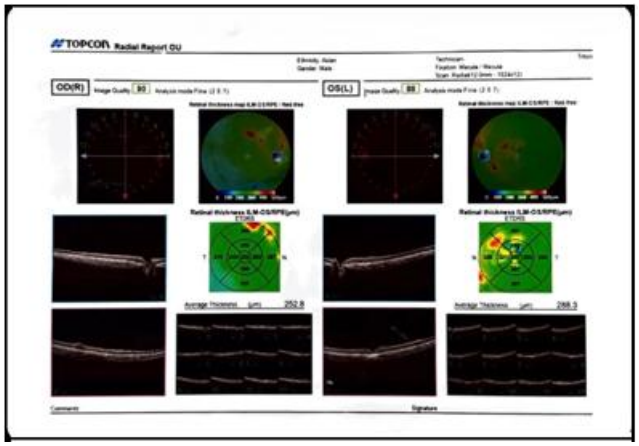
History: A 10-year-old male child presented with slowly progressive, painless diminution of vision in both eyes for three years. There was no history of trauma, night blindness, or systemic illness. Family history was significant for similar visual impairment in the maternal grandfather and a first cousin, strongly suggesting an X-linked inheritance pattern.

Examination: Ocular examination showed normal anterior segment and intraocular pressure. Fundus findings were subtle, showing faint radiating striae at the macula.



Imaging: Optical coherence tomography (OCT) revealed pathognomonic intraretinal schitic cavities involving the inner nuclear and outer plexiform layers in the macular region of both eyes. Based on the clinical triad of male gender, positive family history, and characteristic imaging, a diagnosis of bilateral XLRS was confirmed. The patient was managed conservatively with regular follow-up.





4. Discussion

The diagnosis of XLRS in this 10-year-old patient is supported by the classic presentation of early-onset bilateral vision loss and maternal inheritance. The retinoschisin protein is essential for the structural integrity of the retina; its absence leads to the schisis cavities seen on OCT[1,2].

Table 1: Optical Coherence Tomography features of X-Linked Juvenile Retinoschisis

Parameter	Present Case	Literature Findings
Age at presentation	10 years	First decade of life
Laterality	Bilateral	Predominantly bilateral
Retinal layers involved	INL and OPL	INL commonly affected
Foveal schisis	Present	>90% cases
Peripheral schisis	Present	50–70% cases
Role of OCT	Primary diagnostic tool	Gold standard imaging modality

As demonstrated in this case, fundus findings can be deceptively subtle in the early stages. OCT serves as a critical diagnostic tool, providing high-resolution cross-sectional images that confirm the split within the retinal layers [3,6]. Management remains largely supportive, focused on low-vision aids and monitoring for sight-threatening complications.

5. Conclusion

XLRS should be suspected in any male child presenting with bilateral visual impairment and a positive maternal family history. OCT is vital for establishing an accurate diagnosis, particularly when clinical signs are minimal. Early detection enables appropriate counselling and long-term monitoring to preserve remaining vision.

6. Future Scope

Gene therapy targeting the RS1 gene is an area of active research and may offer definitive treatment in the future. Improved access to genetic testing will further enhance the precision of diagnosis and carrier identification in affected families.

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