

## Dramatic response to topical dorzolamide in X-linked retinoschisis

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Macular involvement is commonly seen in cases with X-linked retinoschisis (XLRS) which includes foveal schisis and cystic maculopathy. Although no definitive treatment has been described, the use of topical 2% dorzolamide hydrochloride in such cases has shown varied response. We herein report a case of XLRS with foveal schisis showing good response to topical dorzolamide. This case highlights the importance of topical dorzolamide in a patient with XLRS.

**Key words:** Dorzolamide, foveal schisis, X-linked retinoschisis

X-linked retinoschisis (XLRS), first described by Haas, is the most common form of juvenile-onset retinal degeneration in males.<sup>[1]</sup> Foveal schisis is seen in most of the cases presenting around in the second to third decade. The cystic involvement can lead to vision loss with progression to macular atrophy in some. There has been no definitive treatment for the management of such cases and the response to dorzolamide have been variable.<sup>[2,3]</sup> Herein, we report a case of XLRS with foveal schisis managed with topical carbonic anhydrase inhibitor (CAI).

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## Case Report

An 8-year-old boy presented with diminution of vision of 4 months duration. Family and systemic history were unremarkable. The best-corrected visual acuity (BCVA) was 6/24 and 6/18 in the right eye and left eye, respectively. The anterior segments were normal in both eyes. Dilated fundus examination revealed foveal schisis with typical cartwheel pattern [Fig. 1a and b]. There was no peripheral schisis in either eye. Foveal schisis was seen as radial hyper autofluorescent streaks on short-wave autofluorescence [Fig. 1c and d]. The right eye, in addition, revealed a small torpedo-shaped lesion inferior to the fovea [Fig. 1a] that was hypo autofluorescent [Fig. 1c]. Spectral-domain optical coherence tomography (SD-OCT) confirmed foveal schisis in both eyes with the central macular thickness (CMT) of 678  $\mu$  and 603  $\mu$  in the right and left eye, respectively [Fig. 2a and b]. Full-field electroretinogram showed negative b-wave that confirmed the diagnosis of juvenile XLRS.

After informed consent, the patient was prescribed 2% dorzolamide hydrochloride eye drops in thrice-daily dosing. At 1-month follow-up, BCVA improved to 6/18 in the right eye and 6/12 in the left eye. SD-OCT showed a drastic reduction in the foveal schisis and CMT decreased to 128  $\mu$  in the right and 135  $\mu$  in the left eye [Fig. 2c and d]. The patient was then advised for regular follow-up.

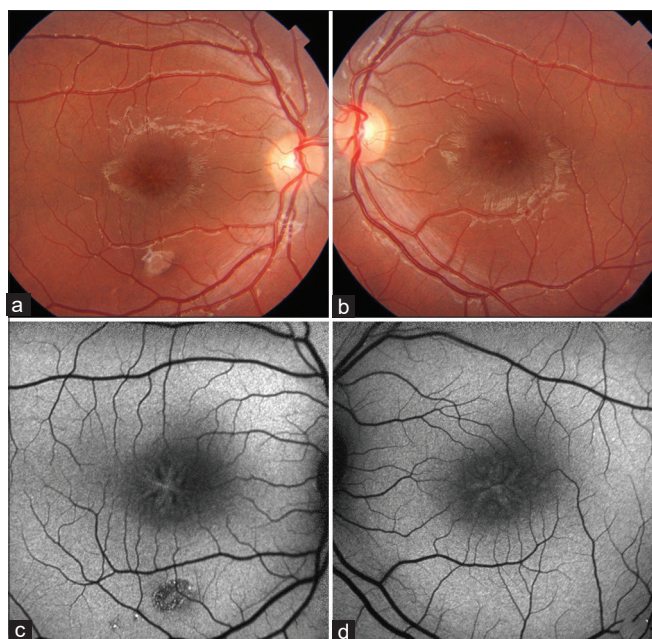
## Discussion

XLRS is caused by the mutations of the *RS1* gene involving Xp22.1 chromosome which is expressed in the photoreceptors and retinal bipolar cells and encodes the secretory protein complex retinoschisin. Retinoschisin is thought to have cell adhesion function and helps in the structural integrity and regulation of cellular fluid balance. Mutations in the *RS1* gene can lead to the absence of retinoschisin protein secretion, secretion of the nonfunctional protein, or decreased

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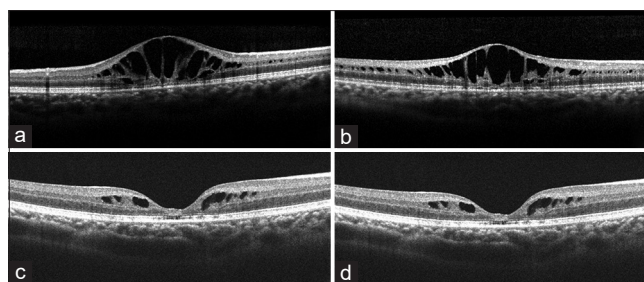


**Figure 1:** Clinical fundus photographs showing the cart-wheel appearance of foveal schisis in (a) the right eye and the left eye (b) autofluorescence imaging (c and d) shows radial hyperautofluorescent streaks corresponding to the schitic cavities

expression of protein resulting in XLRS.<sup>[4,5]</sup> Typical spoke wheel appearance of the foveoschisis is seen on clinical image and FAF. FAF shows the lipofuscin distribution in the RPE and is usually hypoautofluorescent at the macula due to the macular pigments. In foveal schisis, the cystic spaces alter the distribution of the macular pigments, thereby leading to increased transmission of normal background autofluorescence in a radial pattern.

Foveal schisis, a characteristic sign of XLRS, can present in first to the second decade.<sup>[1]</sup> There is no definitive treatment of fovea schisis in the setting of XLRS. Spontaneous resolution may occur but can lead to macular atrophy in some cases, which may further lead to significant vision loss. Treatment modalities used include topical CAI, laser photocoagulation, and advanced therapy such as *RS1* gene therapy. Studies have noted some positive effect of topical dorzolamide on visual acuity, cystoid macular lesions, and central foveal thickness in XLRS and were seen to be independent of the mechanism of dysfunction of retinoschisin. Mild decrease in mid-peripheral schisis and variable response with modest improvement of visual acuity has been seen with CAI.<sup>[2,3,6]</sup> In addition, CAI causes a change in the extracellular pH enhancing the fluid transporter in the RPE cells and thus improves the retinal adhesiveness.

In our case, a drastic response to topical dorzolamide with reduction of cystic macular spaces and improvement of visual acuity was observed within 1 month. This dramatic response of cystic spaces from 678 and 603  $\mu$  to 128 and 135  $\mu$ , respectively on dorzolamide was unusual. The hypoautofluorescent lesion seen in the inferotemporal area



**Figure 2:** Spectral-domain optical coherence tomography (SD-OCT) of both the eyes before (a and b) and after (c and d) 1 month of dorzolamide therapy showing marked reduction in the height of schisis

was an incidental finding which is likely to be a torpedo lesion at an unusual location.

## Conclusion

Early treatment with topical dorzolamide in XLRS can reduce the macular schisis and associated complications such as lamellar and full-thickness macular holes, which can worsen visual acuity at a younger age. Thus, the importance of topical dorzolamide in selected patients with XLRS is highlighted.

## 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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## Conflicts of interest

There are no conflicts of interest.

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