

Surgical management of a large retinal cyst in X-linked retinoschisis with internal drainage: Report of an unusual case

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Although X-linked retinoschisis is a common retinal degeneration condition, the presence of a large retinal cyst obscuring the visual axis in an infant is a rare presentation. Herein, we describe such a case of a child who presented to us with the diagnosis of retinal detachment in both the eyes. However, following multimodal imaging and electrophysiology, the child was found to have bilateral juvenile retinoschisis with a large retinoschisis cyst involving the visual axis seen intraoperatively in the left eye. A limbal approach followed by lensectomy was used to excise the inner retinal layer of the cyst. The intracystic fluid was then drained and the stretched retinal vessels were endocauterized and severed without causing any iatrogenic outer retinal breaks and retinal detachment. The correct diagnosis and meticulous preoperative planning of the surgical procedure helped us manage this challenging case with a favorable anatomical and functional outcome.

Key words: Fovealschisis, Retinal cyst, Retinoschisis, X-linked juvenile retinoschisis

X-linked retinoschisis (XLRs) is a retinal dystrophy caused by the mutation in the RS1 gene and results in schisis or splitting of the inner layers of the retina.^[1] This is in contrast to retinal detachment in which the separation is between the neurosensory retina and retinal pigment epithelium (RPE). This entity has been described in the literature as early as the nineteenth century and with various names, such as “congenital vascular veils,” “juvenile retinoschisis” or “X-linked retinoschisis.”

Case Report

A one-year old boy diagnosed elsewhere as bilateral retinal detachment (RD) secondary to retinopathy of prematurity presented to us with the parents giving a history of squinting in the left eye for the past 5 months. The child was born at

37 weeks of gestation with a birth weight of about 2.5 kilograms. There was no associated history of being in an incubator or postnatal oxygenation or phototherapy. The developmental milestones were normal. The child was able to follow and fixate objects with the right eye but did not follow light with poor fixation in the left eye. Upon examination, the anterior segment of the right eye was found normal with the left eye showing retrolental membrane and a left exotropia. The child was not cooperative for a detailed examination, though the fundus examination of the right eye did raise a suspicion of an inferior retinal elevation [Fig. 1a]. In the left eye, no retinal details were visible other than the presence of a vascular membrane lying behind the clear lens [Fig. 2c]. Ultrasonography also suggested the presence of a moderately reflective membrane in the right eye [Fig. 1b], along with moderately reflective superior and inferior membrane bands in the left eye [Fig. 1c].

The child was examined under general anesthesia. Fundus examination of the right eye (Retcam) now revealed distinct fovealschisis with inferior peripheral retinal schisis [Fig. 1a]. The dark-adapted electroretinogram (ERG) [Handheld ERG Roland Consult Electrophysiology and Imaging (Germany)] showed reduced b-wave amplitude when compared to that of the a-wave; the photopic responses were slightly subnormal [Fig. 2a and b]. Left eye ERG was nonrecordable. Handheld optical coherence tomography (Biotigen, Lieca microsystems) of the right eye revealed the presence of typical foveal retinoschisis [Fig. 1d]. Now, with the diagnosis of XLRs in mind, the left eye was suspected to have a large retinal cyst secondary to a retinal split; a presentation rarely seen in severe cases of XLRs in the very young. The child was taken up for lensectomy with cyst excision.

A 25G trocar-cannula system was used for making the ports. Since the retinal cyst was retrolental, lensectomy was necessary to approach the cyst. In addition, because the inner retinal layer of the cyst was pulled up right behind the lens, the entry of 25G instruments were made directly through the limbus to avoid subretinal entry [Fig. 2c and Video 1]. Infusion was maintained using an anterior chamber maintainer. After lensectomy, retinotomy through the inner retinal layers was performed. Large amounts of altered liquefied blood, indicative of an old standing hemorrhage was drained. After drainage of blood, long slender stretched retinal blood vessels were observed traversing from the inner to the outer retinal layers, which were endocauterized before trimming [Fig. 3a-d and Video 2]. A careful inspection confirmed that there were no outer retinal layer breaks. Therefore, endotamponade was not needed. The limbal ports were sutured by 10-0 VICRYL suture.

The child was reviewed after 1 month and 5 months postsurgery with a stable retina in the right eye and an attached outer retina in the left eye [Fig. 2d and e] with a cycloplegic

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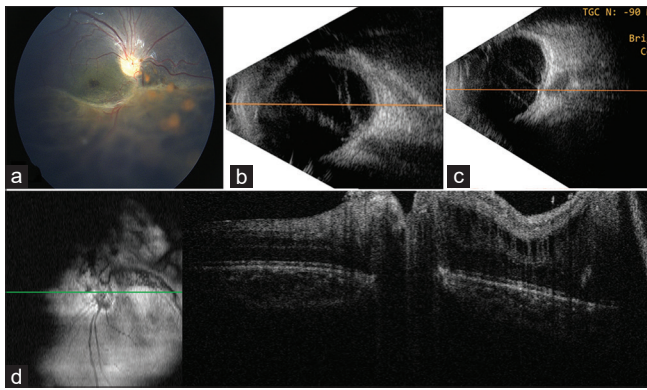


Figure 1: (a) Color fundus photograph (Retcam Image) of the right eye taken under general anaesthesia showing large inferior schisis. (b) Ultrasonography of the right eye showing moderately reflective membrane echo attached at the optic nerve head. (c) Ultrasonography of the left eye showing moderately reflective tractional bands passing antero-posteriorly. (d) Hand held optical coherence tomography (Bioptigen, Lieca microsystems) of the right eye (done under general anaesthesia) showing the schitic retina with the horizontal line scan (Green) passing through the optic nerve head

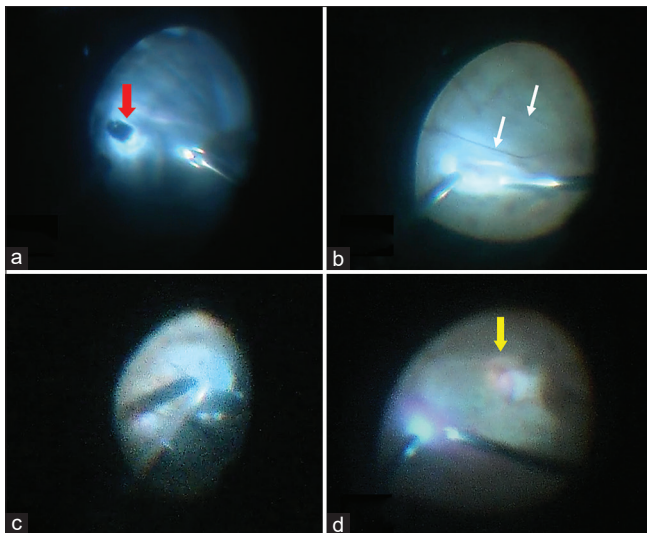


Figure 3: (a) Intraoperative image of the left eye showing the retinotomy (Red arrow) and subsequent drainage of the intraretinal fluid. (b) Transillumination with the help of the endoilluminator shows the retinal vessels traversing across (White arrows). (c) Intraoperative image of the left eye showing endocautery being done to the active bleeders. (d) The optic nerve head (Yellow arrow) could be seen towards the conclusion of the surgery

refraction of (+0.75 spherical/−1.75 cylindrical 180° in the right eye and +15.50 spherical in the left eye). The child was able to follow and fixate objects with the left eye and was advised for patching for further improvement.

Discussion

XLRS is an X-linked inherited retinal degenerative condition, which is characterized by the splitting of the inner retinal layers. The prevalence ranges between 1:5000 and 1:20,000.^[2] Prenner *et al.* classified XLRS into four types based on clinical and OCT findings.^[3] Type 1 had clinically evident foveal schisis, type 2 had macular lamellar schisis, type 3 had additional peripheral

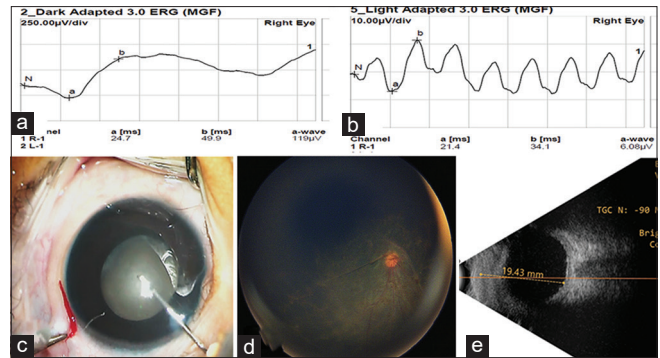


Figure 2: (a) Dark adapted (Scotopic) electroretinogram (ERG) showed a reduction of b-wave amplitude as compared to the a-wave. (b) Subnormal Light adapted (Photopic) ERG. (c) Anterior segment of the left eye at presentation showing the retina behind the clear lens (Photograph taken intraoperatively). (d) Color fundus photograph (Retcam) of the left eye postoperatively with attached outer retina and complete collapse of the large schitic cyst. (e) Ultrasonography of the left eye postoperatively with attached retina

schisis, and type 4 is a combination of types 1 and 3. Although the presence of a negative waveform in ERG is a typical feature of XLRS, advanced cases with large areas of retinal split may have nonrecordable waveforms.^[4] Retinal complications due to XLRS have been documented in the very first year of life and, therefore, timely management becomes crucial.^[5] Nearly half of all the cases show an inferotemporal peripheral schisis with vitreous hemorrhages and/or retinal detachments due to unsupported retinal vessels in about 5% as was seen in our case as well.^[6]

Bullous retinoschisis cavity overhanging the macula is seen in about 8–16% of patients who have congenital XLRS and are below 10 years of age.^[7] Wood *et al.* found long-term stability in 83% of eyes with conversion or progression to a more severe phenotype in the remaining cases.^[8] Rhegmatogenous or combined tractional-rhegmatogenous detachments and vitreous hemorrhage are few complications that require prompt surgical management.

Though uncommon, there are many instances in the literature where faulty diagnoses of rare retinal pathologies have led to disastrous management. Zimmerman and Spencer reported a couple of cases where enucleation was performed on the basis of suspected malignant melanoma of choroid, which were diagnosed to be retinoschisis later.^[9] Ruiz reported enucleation of a blind eye with long standing retinal detachment which was diagnosed clinically as malignant melanoma, later found to be a macrocyst of the retina with organized hemorrhage.^[10] This case report aims to highlight the unusual presentation of retinoschisis as a large retinal cyst which could very well be misdiagnosed as a retinal detachment. A detailed examination under anesthesia with well-planned investigations allowed us to make an accurate diagnosis and a simple surgical plan aimed at draining the cyst led to a successful anatomic and visual outcome.

These large cysts as seen in the very young can be amblyogenic. The drainage of these cysts has been described to collapse the schitic cavity and clear the visual axis, thus preventing amblyopia in children.^[11] Gopal *et al.* have described successful collapse with argon laser photocoagulation.^[12] External drainage of the retinoschitic cysts with indirect visualization has also been described.^[11] However, this was not possible in our case

because the retina was seen just behind the lens with no view of other details because of the intracystic hemorrhage. A more invasive pars plana vitrectomy with entry of instrumentation through pars plana or pars plicata would have led to subretinal infusion of fluid or outer retinal break formation and would have been disastrous.

Conclusion

A high degree of clinical suspicion combined with careful fundus examination, a judicious use of multimodal imaging and electrophysiology under general anesthesia followed by a well-planned surgical procedure helped us achieve a favorable anatomical and functional outcome in an infant with a large retinal cyst with X-linked retinoschisis.

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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