X-Linked Juvenile Retinoschisis Presenting with Rhegmatogenous Retinal Detachment in a Male Nigerian Adolescent: A Case Report

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Abstract

A 15-year-old boy came to the eye clinic with reduced vision in the left eye of a year's duration and prior trauma. Best-corrected visual acuity was 6/9 and hand movement in both eyes, respectively. The anterior segment examination was essentially normal except for a Marcus Gunn pupil and a polar cataract in the left eye. Goldmann applanation tonometry was 10 and 06 mmHg, respectively, in the right and left eyes. Binocular indirect ophthalmoscopy of the right eye revealed pink disc, normal vessels and the Mizuo–Nakamura phenomenon with a cartwheel appearance at the macula. The left eye had a total retinal detachment with proliferative vitreoretinopathy and retinal tear at 12 o' clock. Optical coherence tomography revealed posterior vitreous detachment and schitic cavities at the macula in the left eye. A diagnosis of left rhegmatogenous retinal detachment with background X-linked juvenile retinoschisis was made. The patient was advised on a pars plana vitrectomy under guarded visual prognosis.

Keywords: Cartwheel appearance, Mizuo–Nakamura phenomenon, rhegmatogenous retinal detachment, schitic cavities, X-linked juvenile retinoschisis

Introduction

X-linked juvenile retinoschisis (XJR) is an uncommon condition that is present in males of all races and is a known cause of juvenile macular degeneration.[1] It was first described by Haas in 1898 who laid emphasis on the characteristic radiating maculopathy of XJR.[2] As the name implies, it is a genetic disease due to mutation of the RS-1 gene with locus for inheritance on the Xp22 region of the X chromosome and a paucity of any pathognostic features in female carriers.[3] The clinical features and presentation of XJR vary from reading difficulties to foveal schisis, bullous retinal detachment, vitreous haemorrhage, secondary optic atrophy, neovascular glaucoma, vitreous veils and pigment demarcation lines usually in males of school age.[1,4] Squint and nystagmus may also be unusual features, especially in infants.[5]

The most common feature of XJR is foveal schisis which occurs in about 98%–100% of patients; some other studies have propounded that only 70% of XJR patients have this characteristic schitic lesions though other macula pathology such as pigmentary and atrophic changes of the retinal pigment epithelium may be present, especially in older individuals.[4,6] The Mizuo–Nakamura phenomenon occurs when there is a disappearance of a golden metallic reflex after dark adaptation. This phenomenon is typical of Oguchi disease and may occur in X-linked cone dystrophy.[7] This reflex is believed to occur due to the ineffectiveness of Muller cells in clearing the surge of extracellular potassium ensuing after contact with light.[8]

Case Report

A 15-year-old boy was seen at the eye clinic of the University College Hospital, Ibadan on the 1 November 2021 with a complaint of reduced vision in the left eye following trauma 24 months before presentation. He had been referred from a private facility for left scleral buckling surgery on account of

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a retinal detachment. He had sustained a missile stick injury to the right eye while playing at school 1 year before the onset of visual loss. There was a positive history of pain, tearing and redness at the onset which was managed at an eye clinic in their locality. The patient and guardian said vision was fine after the initial treatment until a year ago. There was no history of flashes, floaters, visual field loss nor night blindness. The patient has never worn spectacles. He is a known asthmatic and haemoglobin genotype is AA. There is no family history of consanguinity, similar symptoms or blinding eye disease.

On ocular examination, the best-corrected visual acuity at presentation was 6/9 and hand movement, respectively, in the right and left eyes. The anterior segment in the right eye was unremarkable, while the left eye had a relative afferent pupillary defect, leucocoria and a polar cataract. The intraocular pressures with Goldmann applanation tonometry were 10 and 06 mmHg in both eyes. Binocular indirect ophthalmoscopy and slit-lamp biomicroscopy with 78 dioptre revealed a right pink disc with a cup disc ratio of 0.3, shiny, golden, metallic glistening reflex at the retina which disappeared with dark adaptation in keeping with Mizuo–Nakamura phenomenon and spoke-like, radiating cystic lesions at the macula in a cart-wheel pattern [Figure 1a]. No areas of peripheral retinoschisis or Seagull appearance were present in the other eye.

An old retinal detachment with the narrow funnel and a retinal tear at 12 o' clock was seen in the left [Figure 1b]. Areas of scarring were observed around the retinal break and inferior retina and fibrovascular proliferation at 4 o' clock with an area of haemorrhage peripherally. Yellowish subretinal exudates were also noticed in the left eye. Spectral-domain optical coherence tomography (OCT) scan of the right eye showed a central foveal thickness of 342 *u*M, a posterior vitreous detachment and schitic cavities of the innermost layers of the retina more pronounced at fovea centralis in keeping with XJR; scans could not be captured for the left eye as the patient could not focus adequately due to the poor vision. The right eye was hyperopic with a refraction of + 0.75DS-0.50DC axis 5°.

A diagnosis of XJR and a left long-standing rhegmatogenous retinal detachment was made based on clinical and optical coherence tomographic scans findings [Figure 2]. Due to the clinical findings, our patient was advised on the need for a left pars plana vitrectomy with perfluorocarbon, silicone oil and endolaser under very guarded visual prognosis due to the proliferative vitreoretinopathy changes. The patient is the first of three children; his siblings, a boy and a girl have no ocular complaints. The guardian has been counselled and advised on the need for a detailed ocular examination for the parents and siblings as regards the incidental finding of X-linked retinoschisis, a hereditary condition in the index patient.

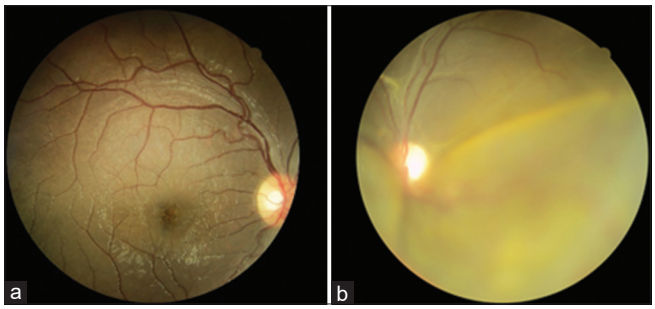

Figure 1: (a) Is the fundus photograph of the right eye showing the glistening sheen in keeping with Mizuo–Nakamura phenomenon and cart-wheel radiation at the fovea while (b) shows a rhegmatogenous retinal detachment with yellowish subretinal exudates and areas of scarring superiorly

Discussion

Our index patient presented with a history of ocular trauma to the left eye with no prior ocular symptoms. XJR was an incidental finding in the right eye diagnosed due to the Mizuo–Nakamura phenomenon, cartwheel appearance at the fovea seen on ophthalmoscopy and schitic cavities on the OCT scan on the right.

XJR is a rare condition and a common aetiology of juvenile macular degeneration.[1] Cases reported from Africa previously include young males of African ethnicity from Nigeria, Mali and South Africa who presented with steady deterioration in vision at the ages of 11, 12 and 19 years, respectively.[9-11] None of these cases presented with retinal detachment and vitreous haemorrhage, but all three patients had the typical radial maculopathy and foveal schisis. It is not definite if the ocular trauma was a red herring or the retinal detachment was secondary to the background XJR as characteristic findings of XJR such as subretinal exudates, neovascularisation and haemorrhage were also present in the affected left eye of our patient.[5,12,13] Approximately 20% of patients with XJR are expected to develop a retinal detachment.[6] The most common devastating ocular complications are vitreous haemorrhage and retinal detachment.[1] These two complications were observed in the complicated eye of this index case.

Spoke-wheel radiations were seen in the right eye with the undetached retina. OCT scan showed schitic cavities in his right fovea. Scans could not be obtained for the left eye because of the patients' inability to fixate due to the poor vision in that eye. Differentials for schitic cavities at the fovea considered in this patient include congenital stationary night blindness and Goldmann–Favre vitreoretinal degeneration. Nyctalopia is a major feature in these two entities and as our patient did not have a history of nyctalopia, they were both ruled out.

Peripheral retinoschisis occurs in about half of patients with XJRS. This was not present in the right eye which had no retinal detachment.[4,6] This may be because a complete posterior vitreous detachment had already taken place in that right eye as captured on the OCT scan as schisis of the retina in XJR is predominantly based on dynamics of the vitreous.[4,14] Refractive errors occur commonly in XJR with hypermetropia being the most prevalent. Our patient was found to be hyperopic after retinoscopy and subjective refraction though he had never worn spectacles in the past.[5] In XJR, the

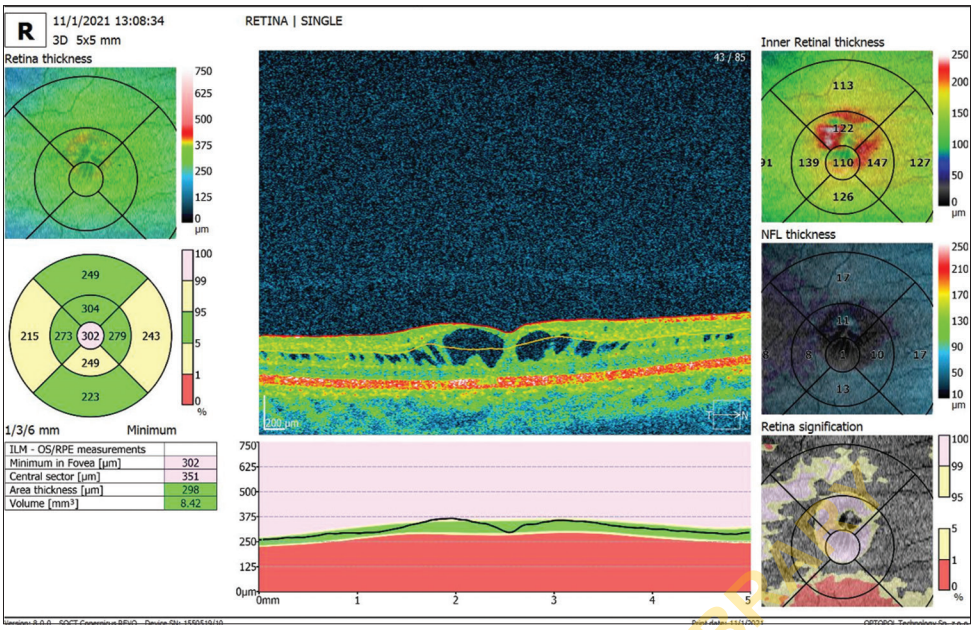

Figure 2: Is the optical coherence tomography scan of the right eye showing schitic cavities along the retina more pronounced at the fovea with a complete posterior vitreous detachment and early disruption of the ellipsoid zone

typical electroretinography finding is the characteristic b-wave reduction.[4,15] This would have further assisted in confirming the diagnosis of XJR, but ERG is unavailable in our facility.

Genetic screening also plays a role in the diagnosis of XJR and ruling out close differentials such as Oguchi disease, Goldman–Favre disease and X-linked cone dystrophy as various genetic mutations of the RS 1 gene has been described.[2,11] This would also have been beneficial in identifying family members with the implicated gene. Genetic analysis was not performed in our patients due to the dearth of these studies in our environment. Our male patient had the distinctive clinical signs of XJR which was relatively sufficient to clinch the diagnosis of XJR.[2,11]

Conclusion

XJR though rare may be the aetiology of poor vision in young males. The presentation may be with complications of retinal detachment as seen in this patient. A detailed clinical examination and high index of suspicion is essential in making a definitive diagnosis after exclusion of differential diagnosis such as Oguchi disease, Goldmann–Favre disease and X-linked cone dystrophy.

Declaration of patient consent

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

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Nil.

Conflicts of interest

There are no conflicts of interest.

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