Juvenile retinoschisis in a 4 year old boy: case report

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ABSTRACT

X-Linked Juvenile Retinoschisis (XLJR) is a rare congenital disease of the retina in children with a prevalence of between 15,000 to 30,000. It almost always occurs in males and is characterized by poor vision due to resultant macular degeneration. We present a case report of this condition in a 4 year old boy whose only presenting symptom was poor vision. He had concurrent mixed astigmatism which initially masked this diagnosis. Failure of the poor vision to improve despite spectacle correction prompted further evaluation. The definitive diagnosis was made through ocular imaging using retinal photography and Optical Coherence Tomography (OCT). High index of suspicion and close follow-up is critical to point to rare cases that need further investigation in unexplained visual symptoms in children; which can be masked by common conditions such as refractive errors and amblyopia.

Key words: Juvenile retinoschisis, X- linked retinoschisis, Juvenile foveoschisis, Juvenile macula degeneration, Poor vision in children

INTRODUCTION

Juvenile retinoschisisis caused by mutations in the RS1 gene, which encodes retinoschisin, a protein involved in intercellular adhesion and likely retinal cellular organization[1]. It is present at birth but usually present at school age due to increased visual demands. The purpose of the case report is to highlight this rare retinal pathology in the paediatric population, the challenges of making the diagnosis in our set-up and to emphasize the importance of patient follow up.

CASE REPORT

This is a case report of a 4 year old male child who presented at a paediatric eye centre in Nairobi with poor vision at both near and far distance for unspecified long duration. He had difficulties in reading and writing at school. There was no variability between day and night vision nor any associated ocular or systemic symptoms.

Initial evaluation of the child revealed visual acuity of 6/36 in both eyes. Cycloplegic refraction confirmed significant mixed astigmatism of +3.00/ -3.00 x180 in both eyes. He had normal anterior segment and unremarkable fundus findings on indirect ophthalmoscopy in both eyes. Spectacle prescription was given. On the first follow-up review after 3 months, the child was still struggling to read at both near and far distance and the visual acuity had only improved by one line to 6/24, despite the spectacle prescription and reported compliance. We queried amblyopia from the uncorrected astigmatism but still ordered further ophthalmological work up because of the marked straining that was noted on attempt to read near work. Corneal topography to assess for keratoconus was normal. Positive findings were found in the retina in both eyes, with fundus photography showing characteristic spoke wheel pattern radiating from the fovea, typical of juvenile retinoschisis (Figure 1a and 1b). Optical coherence tomography also revealed characteristic foveal schisis in juvenile retinischisis involving the superficial neural retina and thinning of the retina (Figure 2a and 2b). The child was immediately put on topical dorzolamide and referred for low vision management. He was advised on environment modification at home and school in addition to the spectacle correction in order to optimize the visual function. He is also on close follow up with the paediatric ophthalmologist and the retina specialist for close to two years since the diagnosis. The vision has remained the same and the serial fundus and optical tomography images have remained stable as per the latest review. The authors have received consent from the parents of the child and permission to publish the case report from Eagle Eye Diagnostic & Laser Centre, Nairobi, Kenya where the patient is being managed.

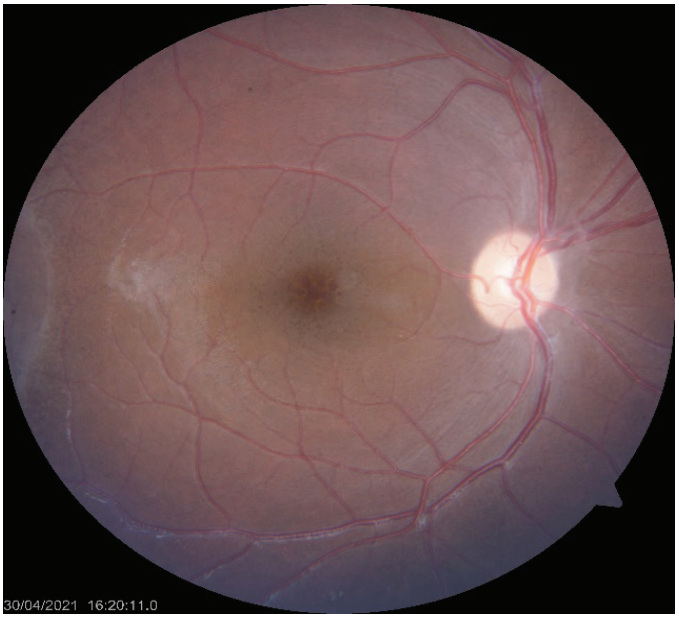

Figure 1a: Fundus photo right eye

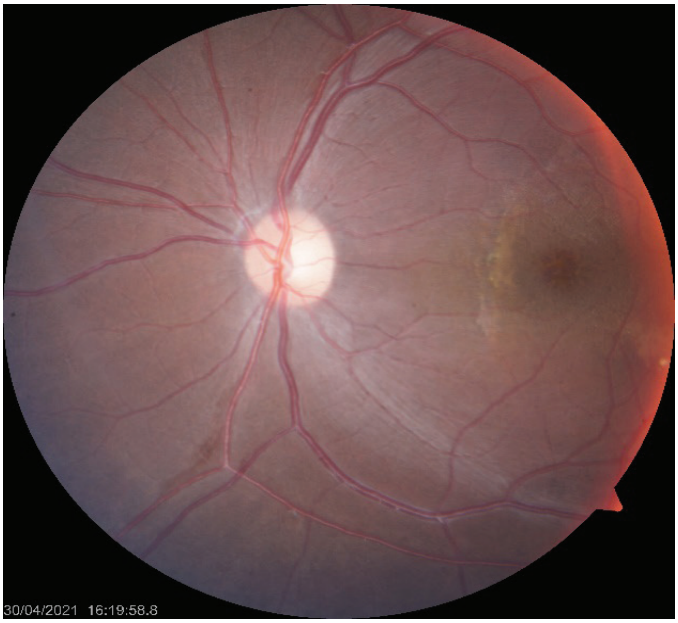

Figure 1b: Fundus photo left eye

Figure 1a and 1b: Characteristic spoke wheel pattern radiating from the fovea for the right and left eye respectively

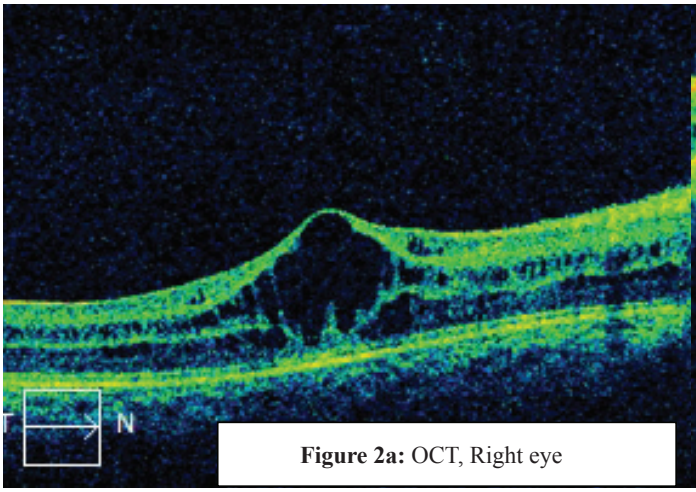

Figure 2a: OCT, Right eye

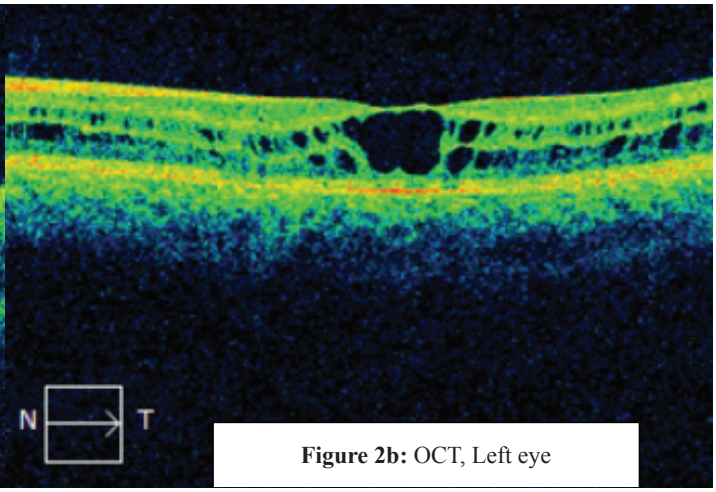

Figure 2b: OCT, Left eye

Figure 2a and 2b: OCT: Cystic spaces primarily in the inner nuclear and outer plexiform layers in the right and left eye respectively

DISCUSSION

Juvenile retinoschisis is one of the main causes of juvenile macular degeneration in males. It mostly presents in school going children with poor vision, as was the case in our patient. Rarely, may it occur in younger children causing nystagmus and strabismus[2].

In childhood the findings can be very subtle, and the diagnosis can easily be missed or confused with amblyopia. The best way to view the characteristic features is by use of a slit- lamp, contact lens and the red free filter[1]. This could explain why it was not possible to pick the findings during the initial clinical fundus examination which was performed using indirect ophthalmoscope and a 20 dioptre lens. In addition, picking the fine retinal changes in a child can be challenging. The characteristic symmetric spoke wheel pattern radiating from the fovea is seen on fundus examination and photography[3].

The criteria used for the clinical diagnosis in absence of genetic testing in our patient were; male patient, presentation in the first decade, characteristic symmetric macular involvement on digital photography and hypo reflective cavities on Optical Coherent Tomography (OCT). OCT is particularly useful in detection of the lamellar schisis which are normally not visible on clinical examination. It also enables detection of splitting of other layers of the retina in addition to the nerve fibre layer. Peripheral retinoschisis occurs in 50% of the patients[4,5]. This had not been observed in this patient as yet. Other useful tests that can be performed to support the diagnosis include; Fundus autoflorescence which reveals increased autoflorescence that highlight the areas of foveal schisis. Fluorescein angiography shows non- petaloid leakage unlike that seen in cystoid macula oedema, with pooling of dye in schisis cavities at late phase. On standard full-field electroretinogram (ffERG) there is reduced b-wave

with preserved a-wave (negative waveform). This ERG finding is not specific to XLJR and can also be observed in other retinal diseases. Genetic testing to confirm the RS1 mutation can be done[1]. The genetic testing was not feasible in our patient.

Management of this condition is by a retinal specialist and/ or paediatric ophthalmologist. Oral or topical carbonic anhydrase inhibitors such as acetazolamide and dorzolamide are effective in flattening the cysts[6-8]. Eventually the cysts gradually reduce by adolescence in the absence of any intervention[3]. Laser photocoagulation to prevent retinal detachment is recommended, though it has a low risk of inducing the same. Low vision devices and environment adjustments such as appropriate sitting position in class, increasing contrast in the text are useful in enhancing the visual functioning. With the new advances on gene therapy, definitive treatment of the XLRS is possible through gene replacement therapy[9]. Genetic counselling is thus paramount for the patient and the relatives.

Complications that can occur include rhegmatogenous retinal detachment, vitreous haemorrhage and foveal ectopia in approximately 5% of the patients[1,8]. Vision often declines in the first two second decades, then stabilises until the fifth or sixth decade. Further deterioration occurs after this period due to macula atrophy, hence lifelong visual monitoring and rehabilitation is essential[3]. XLRS diagnosed in children is often severe with poor prognosis regardless of the age of the patient at presentation[5].

CONCLUSION

High index of suspicion is needed to diagnose such rare retinal diseases in children, which may be masked by other diagnoses such as amblyopia and refractive errors. Where possible further evaluation should be carried out to investigate unexplained visual symptoms in children.

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