RESEARCH ARTICLE

OCT AS A DIAGNOSTIC TOOL IN X-LINKED RETINOSCHISIS

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ABSTRACT

Spectral domain optical coherence tomography was used to image the maculae of patient who had the diagnosis of X-linked retinoschisis maculopathy. here we present a case of 10year old boy with DOV in both eye and retinoschisis changes on ophthalmoscopy and OCT imaging. same changes present to his brother also with suspected positive history to maternal grandfather at M.G.M. Medical college &MYH Hospital Indore, India.

Key words: x-linked retinoschisis, oct, maculopathy

INTRODUCTION

X-linked retinoschisis (XLRS) is a congenital retinal dystrophy caused by mutations of the RS1 gene on the short arm of the X chromosome, Xp22.1 Nearly all patients with XLRS display maculopathy, which can vary in appearance from foveal radial striations to macular cysts or atrophic lesions. Less than half of patients with XLRS present with peripheral retinoschisis; however, complications, such as retinal detachment and vitreous hemorrhage, are more frequent in that subgroup of patients. Several publications describing the optical coherence tomography (OCT) characteristics of XLRS have noted different findings for this condition. Splitting in the nerve fibre layer (NFL) attributed to Muller cell dysfunction was noted on histological studies and was observed in vivo with OCT by Eriksson et al. Additionally, groups have described splitting in retinal layers deep to the NFL with OCT.

Case Report

A 10year old boy(Fig.1) presented in ophthalmology department M.G.M. Medical college &MYH Hospital Indore, India with gradual, painless and progressive diminution of vision in both eye since (LE>RE) since 6 month. there were no associated symptoms. patient maternal grandfather has severe diminuation of vision since 10yr back and same history of DOV to his brother. there was no any past and personnel history. clinically he was well oriented to time, place and person. on opthalmic evaluation best corrected visual acuity was 6/24 OD and 3/60 OS without improvement on manifest refraction and pin hole. on retinoscopy error was was +1.5 OD and -6.5 OS diopter spherical. Rest Anterior segment within normal limit. On fundus examination of right eye shows hazy media due to vitreous hemmorhage(inferiotemporal ti disc),Absent foveal refax ,elevated macular area with radiating fold and whitish band like elevated membrane with distinct margins seen in the inferotemporal quadrant from 7 to 10 0 clock.[Vitreous Veils?] ,Inner layer shows oval defect on general fundus. left eye shows only Absent foveal reflax, elevated macular area with radiating fold.

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Investigations

- Blood and urine investigations within normal limit.
- OCT – patient (Fig.4,5) and his brother (Fig.6,7) shows cleavage of retina into two planes, which his connected by thin walled vertical palisades, separated by low reflective cystoids spaces, which is more confluent and prominent in foveal region.

DISCUSSION

X-linked juvenile retinoschisis (XLRS) is an inherited early onset retinal degenerative disease and is the leading cause of juvenile macular degeneration in males. Characteristics include moderate to severe loss in central vision caused by schisis (splitting) of the layers of the retina. The disease is transmitted as an X linked recessive trait, occurring almost exclusively in males, although a few affected female carriers have also been identified, some having a family history of consanguinity.\textsuperscript{9,10} Axial hypermetropia also appears to be a consistent feature of XLRS.\textsuperscript{11} The diagnosis of XLRS is based on the findings of fundus examination, electrophysiologic testing and molecular genetic testing. A recent addition to the armamentarium of
useful investigations for XLRS is optical coherence tomography (OCT) (fig 3). This is a non-invasive, non-contact procedure that uses low coherence interferometry to detect relative reflection changes and different optical surfaces. The detail from the SD-OCT scans was ideal for visualizing the retinal architecture and cystic changes in the eyes with XLRS, thereby allowing for precise identification of the pathologic changes and their location within the structure of the retina. The enhanced retinal detail that is possible with SD-OCT is not only beneficial to earlier diagnosis, but may also aid in the previously described classification schemes for XLRS. At present, no treatment to halt the natural progression of schisis formation is available. One recent report describes successful treatment of schisis cavities with topical dorzolamide (carbonic anhydrase inhibitor).

References


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