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Case Report

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JUVENILE RETINOSCHISIS: A CASE REPORT

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ABSTRACT

Juvenile retinoschisis is a hereditary macular dystrophy that is inheritated in the XR (X-linked recessive) manner. Macular folds associated with or without peripheral retinoschisis are major clinical signs, which are responsible for decreased visual acuity. A 17 year old male boy who came in OPD for a consultation for progressive diminution in visual acuity since 5 years. Visual acuity was 3/60 without correction; and the left eye at 3/60, After the myopic correction both eye achieved a 5/60 best corrected visual acuity. The fundus examination revealed supero-nasal juxtamacular radial lines with increased separation with some microcysts in both eyes. Superonasal retinal splitting with microcyst was the major examination finding in retinal periphery.

KEYWORDS: Retinoschisis, Dystrophy, Cart Wheel Hole.

INTRODUCTION

Juvenile retinoschisis is one of the primary causes of macular dystrophy in young boys. It is characterized by a high degree of clinical inconsistency.[1,2] Macular star with or without peripheral retinoschisis are major clinical signs. Separation occurs in the retina, mainly at the nerve fiber layer. It is responsible for a decline in the visual acuity of varying significance and is slowly progressive, generally appearing during the first decade.

No effective treatment to stop the progression of the macular degeneration till now, clinical management of amblyopia and the surgical correction of some complications are the major focusing area of the treatment guideline.[3]

CASE REPORT

This study was regarding a 17-year-old male boy who came in OPD for a consultation for progressive diminution in visual acuity over 5 years. Uncorrected distance visual acuity without correction was at 3/60 in both eyes ; After myopic correction best corrected visual acuity was scored at 5/60 in both eyes. In each eye slit-lamp biomicroscopy examination found a normal anterior segment, normal intraocular pressure, and a clear lens. Indirect gonioscopy found bilateral open angles without any anomalies in both eyes. A vitreous examination was within normal limits. Fundus examination showed perimacular radial lines (like a cart wheel appereance) with increased separation with some microcysts more pronounced in nasal side in both eyes (figure 1,2). Retinal periphery examination of both the eyes found superonasal retinal splitting with a demarcation line corresponding to separation. On optical coherence tomography; cystic spaces was seen in inner layers of retina in both eyes (figure 3,4). Retinal angiography identified perimacular radial lines, with superonasal bilateral peripheral separation. Fluorescein injection did not show macular edema or vascular degeneration. Color vision revealed dyschromatopsia without an axis. An examination of five members of the family revealed a trisomy in the 12-year-old younger brother without the involvement of eye anomalies in parents and siblings. Molecular biological tests could not be performed.

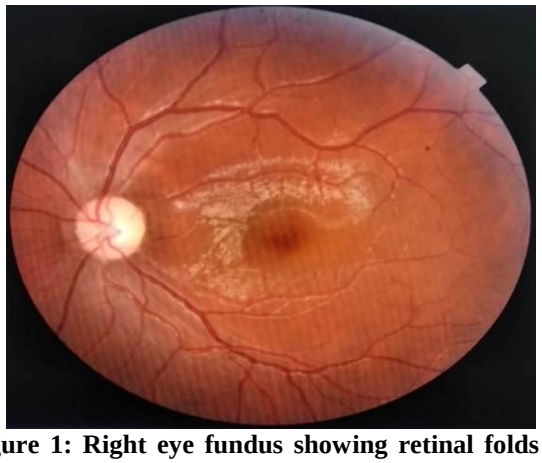

Figure 1: Right eye fundus showing retinal folds on juxta foveal area more pronounce in nasal side.

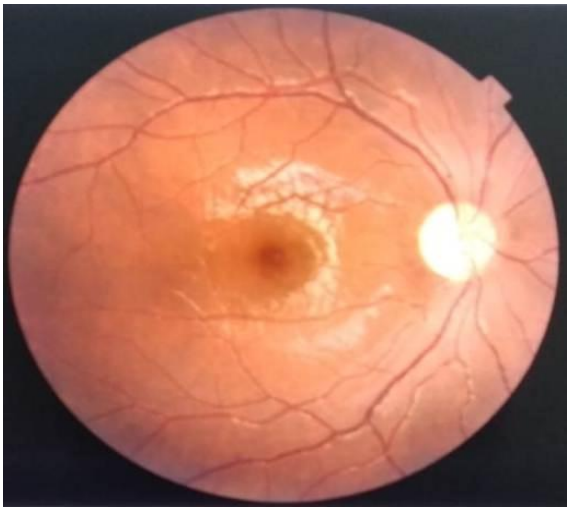

Figure 2: Left eye fundus showing retinal folds in juxtra foveal area more in nasal side of the fovea.

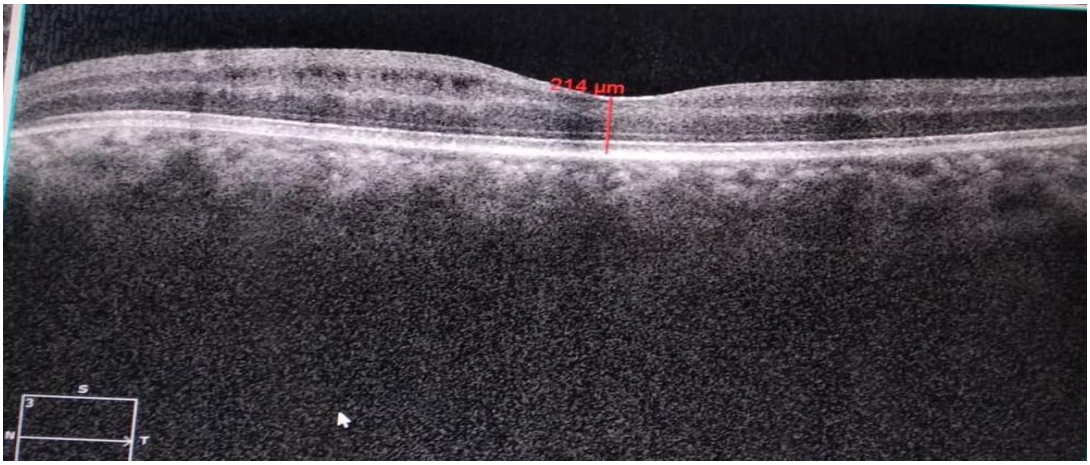

Figure 3: Right eye Optical coherence tomography showing retinal splitting at the level of inner retinal layer with microcysts more in nasal side.

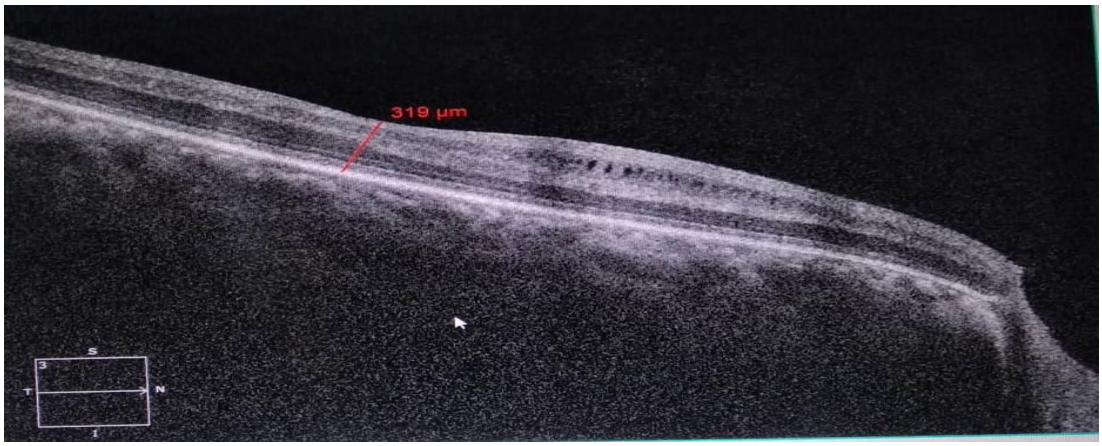

Figure 4: Left eye optical coherence tomography showing retinal splitting at the level of inner layer with microcyst more in nasal side.

DISCUSSION

Juvenile retinoschisis is the most common juvenile macular degeneration disease.[1,2] It only affects boys with variable expressivity. Its global prevalence varies between 1/ 5000 and 1/25,000.[4,5] The highest prevalence has been reported in Finland.[6,7] A decline in visual acuity is commonest clinical symptom involving retinoschisis.[8,10] This decline in visual acuity is generally detected in boys in school going age. Visual acuity at this school going age is extremely unpredictable, but is generally above 6/30. In our case, the visual acuity in the best eye was 3/60 at 12 years old. Hypermetropia is a frequent finding but was not true in

this case. A family examination is crucial. It makes it possible to either clarify the known family history or, conversely, to detect genetic abnormalities in the genology, as was the true for this case. Pathognomonic sign was the macular separation.[9-11] Sometimes, it may be absence of radial fold extending from fovea to macular periphery; and in 50% of the patients, peripheral retinoschisis is inferotemporal, as was not true for our case. Electrophysiological disturbances are related to neuroretinal damage. A possible involvement of foveolar Müller cells in the formation of the idiopathic macular hole has been suggested. This case could, thus, support the hypothesis of the participation of foveolar Müller cells in the pathogenesis of the macular hole.[12]

SUMMARY & CONCLUSION

17 year male boy came to consult his decline in visual acuity, first sight he was diagnosed with simple myopia, but with myopic correction VA was not improved that much.after the through investigation like OCT and fundus angiography. He diagnosed as juvenile retinoschisis.

X-linked juvenile retinoschisis is a condition affecting young boys and is responsible for a visual disability; it may Progress to macular hole which more worsen the visual status of the individual. So regular follow up visits, fundus examination, amblyopic management and genetic counselling is advised for siblings.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the consent form the patient has given his consent for his fundus, OCT image and other clinical information to be reported in the journal.

Financial support

Nil.

Conflicts of interest

There are no conflicts of interest.

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