ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 08 | August - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

CLINICAL PROFILE OF RETINOSCHISIS AT A TERTIARY EYE CARE CENTER IN BUNDELKHAND REGION.

Ophthalomology

Dr. Jitendra Kumar M.S. Ophthalmology, Professor and Head of the Department, Department of Ophthalmology, M.L.B. Medical College, Jhansi (U.P.).

Dr. Ruby Bala Junior Resident, Department of Ophthalmology, M.L.B. Medical College, Jhansi (U.P.)

Dr. Nistha Singh Junior Resident, Department of Ophthalmology, M.L.B. Medical College, Jhansi (U.P.)

ABSTRACT

Purpose: To study the clinical profile of patients with Retinoschisis in Department of Ophthalmology at M.L.B Medical College Jhansi. **Materials and Methods:** Prospective study of 40 patients of Retinoschisis present in Department of Ophthalmology at M.L B Medical College Jhansi. **Results:** A total of 40 patients were examined out of which 22(55%) patients were x-linked and 18(45%) patients were acquired. Maximum no of patients present at the age group of 11-15 years(30%) , followed by16-20 years 10(25%),followed by5-10 years 08(20%) ,followed by 21-25 years 07(17.5%) and least by the 26-30 years (7.5%). All the patients have cystic space in inner nuclear and outer plexiform layer in OCT. In fundus of 22(48%) patient have found peripheral dendritic lesions, 18(45%) patients have found Bicycle wheel like maculopathy, 06(15%) have found typical oval inner veils defect and in 02(5%) have found vitreous veils . **Conclusion:** Most common form of Retinoschisis is inherited form found in male , present bilaterally . Maximum number of patients come at the age group of 11-15 years . Peripheral dendritic lesions is the most common finding in fundus of patients of Retinoschisis and Cystic space in inner nuclear and outer plexiform layer in OCT.

KEYWORDS

X-Linked, Inherited , Retinoschisis , Dendritic ,Maculopathy

INTRODUCTION

X-linked retinoschisis, with a prevalence of about 1 in 15,000 to 30,000, is one of the main causes of juvenile macular degeneration in males.[2] It is characterized by symmetric bilateral macular involvement beginning in the first decade of life.[3] It is caused by a large variety of mutations in the *RS1* gene on Xp22.1-p22.3, which encodes the protein retinoschisin. This protein is involved in intercellular adhesion and likely retinal cellular organization. X-linked retinoschisis is inherited in an X-linked manner with complete penetrance and variable expressivity. Most affected individuals are males, as heterozygous females are rarely affected. However, retinoschisis has been reported in non-consanguinous females.[4][5] The phenotype can be markedly variable even within the same genotype, and can involve the peripheral retina.X-linked retinoschisis is linked to mutations in *RS1* on Xp22.1-p22.3. The gene encodes a 224-amino acid protein called retinoschisin, which is secreted by photoreceptors. [6] This protein is found throughout the retina, and is thought to be involved in cell-cell adhesion and intercellular matrix retinal architecture development through interactions with αβ crystallin and β2-laminin. On histopathological examination, the splitting in X-linked retinoschisis occurs predominantly in the nerve fiber layer. [7] **Senile Retinoschisis**, also called **degenerative retinoschisis** or **acquired retinoschisis**.The prevalence is about 5% of the population over the age of 20 year. This is a microcystoid degeneration of the neurosensory retina, with splitting at the outer plexiform layer of the retina.

MATERIALAND METHOD

This was a Prospective study that involved 80 eyes of 40 patients of Retinoschisis complaining of diminution of vision . Patients were recruited from the OPD of MLB medical Jhansi, Uttar Pradesh and followed from 1st may 2022 to1st March 2023 . It was performed under the Helsinki Declaration of 1975 ,as revised in 2000.The necessary from the Ethical and Research Committe was obtained for the study.

Inclusion Criteria

All patients who presented to the OPD of MLB Medical College Jhansi with the complaint of diminution of vision were included.

Exclusion Criteria

1. Patients with ocular systemic diseases that could affect the retina.
2. Patients with other retinal disorders.
3. Patients with recent intraocular surgery.
4. Patients with the history of trauma.

All patients were subjected to a detailed history taking, refraction using Topcon Autorefractometer and best corrected visual acuity (VA) measurement. All patients had complete ophthalmic examination including Biomicroscopic slit lamp examination, Fundus examination with 90D lens and Fundus photography and Spectral Domain Optical Coherence Tomography.

RESULT

A total of 80 eyes of 40 patients were studied. We included eyes with complain of diminution of vision were included.

Table-1

Age Groups	No Of People
5-10 Y	08(20%)
10-15Y	12(30%)
15-20Y	10(25%)
21-25Y	07(17.5%)
26-30Y	03(7.5%)

Table-2 Fundus Findings In Patients Of Retinoschisis

	FUNDUS FINDINGS
22(48%)	Peripheral dendritic lesion
18(45%)	Bicycle wheel like maculopathy
06(15%)	Large typical oval inner veils defects
02(5%)	Vitreous veils

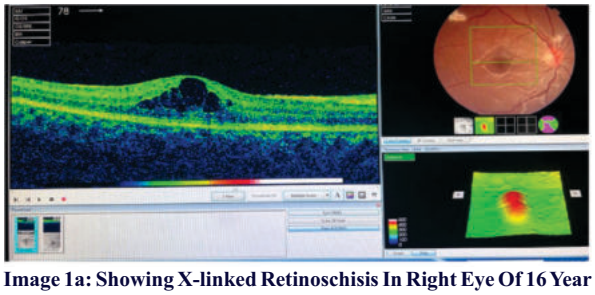

Image 1a: Showing X-linked Retinoschisis In Right Eye Of 16 Year Old Male. Oct Showing Cystic Spaces In Inner Nuclear And Outer Plexiform Layer Involving Rnfl And In Fundus Showing Typical Peripheral Dendritic Lesions And Maculopathy.

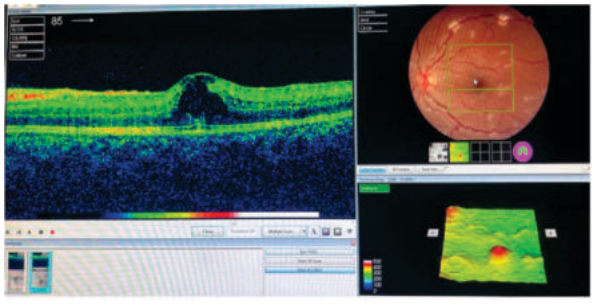

Image 1b: Showing X-linked Retinoschisis In Left Eye Of 16 Year

Old Male. Oct Showing Cystic Spaces In Inner Nuclear And Outer Plexiform Layer Invoiving Rnfl And In Fundus Showing Typical Peripheral Dendritic Lesions And Maculopathy.

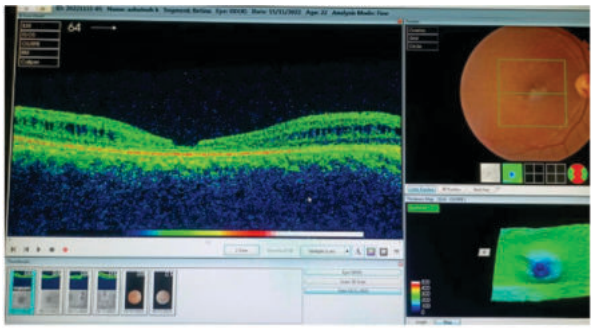

Image 2a: Showing Juvenile Retinoschisis In Right Eye Of 22 Year Old Male. Oct Showing Cystic Spaces In Inner Nuclear And Outer Plexiform Layer , Not Invoiving Rnfl.

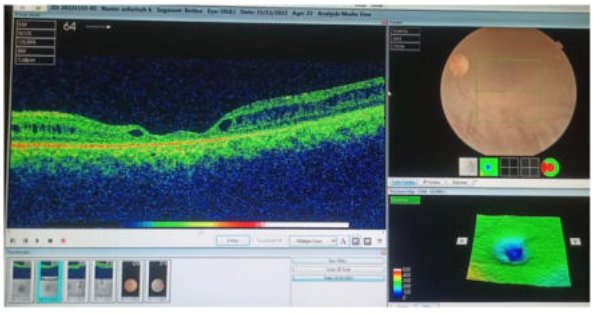

Image 2b: Showing Juvenile Retinoschisis In Left Eye Of 22 Year Old Male. Oct Showing Cystic Spaces In Inner Nuclear And Outer Plexiform Layer , Not Invoiving Rnfl.

DISCUSSION

Future Gene Therapies

As retinoschisin is expressed in the retina during early development and maintained throughout life, gene replacement has become a target for therapeutic intervention. Viral vector delivery of the *RS1* gene in CXLRS mouse models shows promise, as retinoschisin was successfully expressed in all retinal layers and the b-wave amplitude was restored on electroretinography (ERG). [8], [9] Intravitreal injection of adeno-associated viral (AAV) vectors AAV8 and recombinant AAV2 (rAAV2) carrying functional *RS1* into *Rs1* knockout (KO) mouse models has demonstrated effectiveness. These targets are now currently being studied in human clinical trials. In the *Rs1*-KO mice, AAV8-mediated gene delivery induced reorganization of the photoreceptor-depolarizing bipolar cell synapse and was found to restore function. [10]

Currently, there are 2 gene therapy trials underway for the treatment of CXLRS. The National Eye Institute is conducting a phase 1/2 dose-escalation clinical trial for AAV8-scRS/IRBPhRS gene transfer in patients with CXLRS (NCT02317887). A biotechnology company, Applied Genetic Technologies Corporation, is also performing a phase 1/2 dose-escalation clinical trial for a rAAV2tYF-CB-hRS1 therapy (NCT02416622). Although both studies attempt to optimize dose, they are primarily designed to evaluate safety.

REFERENCES

1. American Academy of Ophthalmology. X-linked juvenile retinoschisis. https://www.aao.org/image/x-linked-juvenile-retinoschisis Accessed May 20, 2020.
2. ↑ Sikkink SK, Biswas S, Parry NRA, Stanga PE, Trump D. X-linked retinoschisis: an update. Journal of Medical Genetics. 2007 Apr 1;44(4):225–32.
3. ↑ Jump up to:3.0 3.1 3.2 3.3 3.4 3.5 Sieving PA, MacDonald IM, Hoang S. X-Linked Congenital Retinoschisis. 2003 Oct 24 [Updated 2020 Nov 5]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1222/
4. ↑ Saleheen D, Ali A, Khanum S, Ozair MZ, Zaidi M, Sethi MJ, et al. Molecular analysis of the XLRS1 gene in 4 females affected with X-linked juvenile retinoschisis. Can J Ophthalmol. 2008 Oct;43(5):596–9.
5. ↑ Rodríguez FJ, Rodríguez A, Mendoza-Londoño R, Tamayo ML. X-linked retinoschisis in three females from the same family: a phenotype-genotype correlation. Retina (Philadelphia, Pa). 2005 Jan;25(1):69–74.
6. ↑ Grayson C, Reid SNM, Ellis JA, Rutherford A, Sowden JC, Yates JRW, et al. Retinoschisin, the X-linked retinoschisis protein, is a secreted photoreceptor protein, and is expressed and released by Weri–Rb1 cells. Hum Mol Genet. 2000 Jul 22;9(12):1873–9.
7. ↑ Yassur Y, Nissenkorn I, Ben-Sira I, Kaffe S, Goodman RM. Autosomal Dominant Inheritance of Retinoschisis. American Journal of Ophthalmology. 1982 Sep 1;94(3):338–43.
8. Zeng Y, Takada Y, Kjellstrom S, et al. RS-1 gene delivery to an adult RS1h knockou mouse model restores ERG b-wave with reversal of the electronegative waveform of X-linked retinoschisis. *Invest Ophthalmol Vis Sci.* 2004;45:3279-3285.
9. Min SH, Molday LL, Seeliger MW, et al. Prolonged recovery of retinal structure/function After gene therapy in an RS1h-deficient mouse model of X-linked juvenile retinoschisis. *Mol Ther.* 2005;12:644-651.
10. Ou J, Vijayasarathy C, Ziccardi L, et al. Synaptic pathology and therapeutic repair in adult retinoschisis mouse by AAV-RS1 transfer. *J Clin Invest.* 2015;125:2891-2903.