

## Long Term Follow-Up in a Female Patient with X-Linked Juvenile Retinoschisis Resulted from Consanguineous Marriage

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**Abstract:** Female patients with X-linked juvenile retinoschisis were extremely rare. Over 40 years ago, a female patient from a consanguineous marriage was reported in our institute. We present long term follow-up of this female patient. Our findings may contribute to a better understanding of the natural course of X-linked juvenile retinoschisis in woman.

**Keywords:** X-linked juvenile retinoschisis, consanguineous marriage.

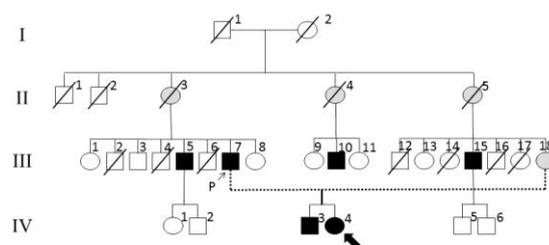
### INTRODUCTION

X-linked juvenile retinoschisis (XLRS) is characterized by vitreous degeneration and splitting of the retina between the nerve fiber and ganglion cell layers. The mode of transmission is X-linked recessive, primarily affecting males. In 2003, Ali *et al.* [1] reported the first case of homozygous XLRS occurrence in females from a consanguineous marriage. Before their report, Uchino and Shimizu [2] described a female

patient in Japan. We present long term follow-up of this patient.

### CASE REPORT

Over 40 years ago, a female patient with XLRS from a consanguineous marriage was reported in our institute [2]. First, their family tree for four generations is shown in Figure 1.



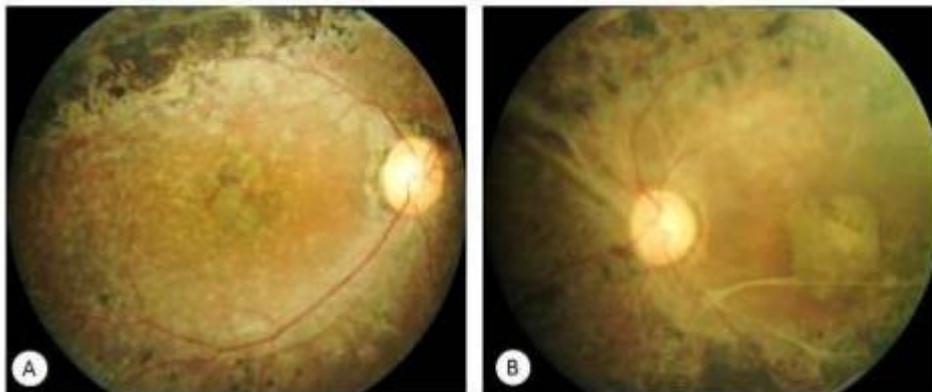
**Fig-1: Pedigree of the affected family. (consanguineous marriage; III-7 and III-18)**

(Open circle: normal woman; black circle: affected woman; open square: normal man; black square: affected man; speckled circle: suspected carrier woman, P: proband, arrow indicates this patient)

This female patient (IV-4 in Figure 1) was diagnosed with XLRS at the age of nine. Her best-corrected visual acuity (BCVA) was 0.15 in the right eye and 0.01 in the left eye. She had typical retinoschisis with macular involvement in both eyes. Her parents were cousin married (III-7 and III-18 in Figure 1). Her brother (IV-3 in Figure 1) was diagnosed with XLRS at the age of 15. His BCVA was 0.2 in the right eye and 0.1 in the left eye. He had typical retinoschisis with macular involvement in both eyes. Her father (III-7 in Figure 1) was diagnosed with XLRS at the age of 48. His BCVA was 0.08 in the right eye and no light perception in the left eye. He had typical retinoschisis with macular involvement in the right eye and retinal detachment in the left eye. Her uncle (III-5 in Figure 1) was diagnosed with XLRS. His BCVA was

0.04 in the right eye and 0.03 in the left eye. He had typical retinoschisis with macular involvement in both eyes. Cousin of her father (III-10 in Figure 1) was suspected with XLRS. Her uncle (III-3 in Figure 1) was clinically normal. Her mother (III-18 in Figure 1) was clinically normal. Her uncle (III-15 in Figure 1) was suspected with XLRS. His BCVA was 0.5 in the right eye and 0.6 in the left eye.

The patient was followed up for 45 years. At the age of 40, her BCVA was 0.03 in the right eye and counting finger in the left eye. Slit-lamp examination revealed nuclear opacity in both lenses. Fundus examination revealed marked chorioretinal atrophy with macular involvement (Figure 2). Moreover, retinal gliosis was also observed in both eyes.

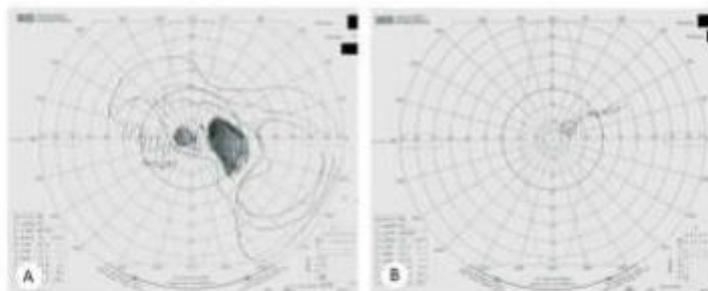


**Fig-2: Fundus photographs of the right (A) and left (B) eyes**

Note marked chorioretinal atrophy with macular involvement in both eyes.

paracentral scotoma in the right eye, while the visual field of the left eye showed marked constriction (Figure 3).

Goldmann perimeter was performed and showed moderate peripheral constriction with



**Fig-3: Goldmann perimetry of the (A) right and (B) left eyes**

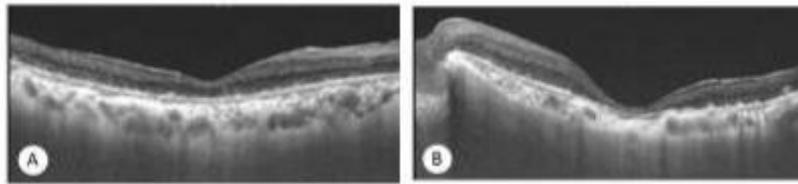
At the age of 54, her BCVA was 0.01 in the right eye and counting finger in the left eye. Nuclear opacity was gradually progressed in both lenses. Fundus

examination showed no remarkable changes; however, details were unclear due to cataract (Figure 4).



**Fig-4: Fundus photographs of the right (A) and left (B) eyes**

Optical coherence tomography (OCT) revealed a widespread disruption of the ellipsoid zone with marked reduced retinal thickness (Figure 5).



**Fig-5:Optical coherence tomography images of the right (A) and left (B) eyes**

Note a widespread disruption of the ellipsoid zone with marked reduced retinal thickness. The patient continues to receive low vision rehabilitation.

## DISCUSSION

Juvenile retinoschisis is transmitted as X-linked recessive inheritance. However, there have been few reports of female patients with this disorder from a consanguineous marriage [2-5]. Forsius *et al.* [3] described a 44-year-old female patient. Her parents were consanguineous marriage. It has been ascertained that all her sons by two different fathers are affected, and that her sister is a conductor, with an affected son. Shimazaki and Matsuhashi [4] reported two female patients, a mother (49-year-old) and daughter (23-year-old), with bilateral foveal changes that resembled those of XLRS. Yamaguchi and Hara [5] reported a 2-year-old girl from a consanguineous marriage. This present patient was the second youngest case of woman with XLRS. Moreover, it was extremely rare that we could observe her for over 40 years. Our findings may contribute to a better understanding of the natural course of XLRS in woman.

## REFERENCES

1. Ali A, Feroze AH, Rizvi ZH, Rehman TU. Consanguineous marriage resulting in homozygous occurrence of X-linked retinoschisis in girls. *Am J Ophthalmol.* 2003; 136(4): 767-769.
2. Uchino M, Shimizu H. A family with congenital retinoschisis including a female patient. *Rinshouganka.* 1976; 30(2): 145-151.
3. Forsius H, Vainio-Mattila B, Eriksson A. X-linked hereditary retinoschisis. *Br J Ophthalmol.* 1962; 46(11): 678-681.
4. Shimazaki J, Matsuhashi M. Familial retinoschisis in female patients. *Doc Ophthalmol.* 1987; 65(3): 393-400.
5. Yamaguchi K, Hara S. Autosomal juvenile retinoschisis without foveal retinoschisis. *Br J Ophthalmol.* 1989; 73(6): 470-473.