Autosomal juvenile retinoschisis without foveal retinoschisis

KATSUHIRO YAMAGUCHI AND SATOSHI HARA

From the Department of Ophthalmology, School of Medicine, Tohoku University, Japan

SUMMARY We describe hereditary retinoschisis without foveal retinoschisis in a girl (the product of a consanguineous marriage) and her father. The father had peripheral retinoschisis with dendritic changes of the retina and good visual acuity. The daughter had peripheral retinoschisis complicated by vitreous haemorrhage and retinal detachment. Neither had foveal retinoschisis. The pattern in a scotopic single-flash electroretinogram differentiated this condition from other forms of retinoschisis. We believe that these cases represent the autosomal recessive form of juvenile retinoschisis without foveal retinoschisis.

Autosomal juvenile retinoschisis is a rare vitreoretinal dystrophy characterised by peripheral retinoschisis without pathognomonic foveal retinoschisis. We describe a father and daughter who had peripheral retinoschisis without foveal retinoschisis, which possibly was transmitted as an autosomal recessive disorder. The results of a scotopic single-flash electroretinogram showed a slightly reduced a wave, a diminished b wave that returned to above baseline levels, and diminished oscillatory potentials, a pattern considered pathognomonic in this disorder.

Case reports

CASE 1
A 2-year-old girl suffered a fall on 16 May 1987. Several days later her mother noticed the presence of right esotropia. The child was referred to Tohoku University Hospital because of a vitreous haemorr-
Autosomal juvenile retinoschisis without foveal retinoschisis

Fig. 2  The left inferotemporal peripheral fundus of the proband shows retinoschisis and a large break in the inner layer of the retina.

Fig. 3  The left macula of the proband appeared normal, and no foveal retinoschisis is observed.

Ultrasoundography disclosed a strong sheet-like echo that was suggestive of a retinal detachment.

After a week’s observation an ophthalmoscopic examination of the girl’s right eye showed a small, full-thickness retinal detachment that involved the inferotemporal retina and extended from the 5 to 9 o’clock meridian. Also observed were a large grey membrane floating in the inferotemporal vitreous cavity and two holes in the outer retinal layer at the 6 o’clock meridian between the vascular arcade and equator, among the old vitreous haemorrhage. Another lesion of retinoschisis spread across the superotemporal fundus. The optic disc appeared normal, and foveal retinoschisis was not observed.

Surgical treatment consisting of cryoretinopexy and scleral buckling was performed successfully on 9 June 1987. The retina became reattached during the postoperative period of observation.
displayed a slightly reduced a wave, a diminished b wave that returned to above baseline levels, and diminished oscillatory potentials (Fig. 6). Fluorescein angiography disclosed arborescent figures of retinal vessels in the regions of retinoschisis.

**Discussion**

Among the various hereditary retinoschisis foveal retinoschisis is an important pathognomonic finding. In X-linked juvenile retinoschisis, which is carried by the X chromosome and is transmitted in a recessive manner with a high degree of penetrance, foveal retinoschisis occurs in 98% to 100% of patients. In approximately 50% of patients foveal retinoschisis is the sole pathological finding. This disease is characterised by a negative electroretinogram, which showed a normal a wave, a b wave that does not return to the baseline values, and reduced oscillatory potentials. In a female carrier of X-linked juvenile retinoschisis radial wrinkling of the inner limiting membrane occurs around the fovea, but no peripheral retinoschisis is seen. The electroretinogram is normal. Favre-Goldmann disease, which is inherited in an autosomal recessive manner, is not only characterised by peripheral retinoschisis and foveal retinoschisis but also includes an optically empty vitreous, chorioretinal atrophy, cataract, loss of peripheral vision, glial bands, eventual blindness, and an extinguished electroretinogram. In autosomal recessive foveal retinoschisis the lesion is limited to the macular area. The electroretinogram is normal. Autosomal dominant juvenile retinoschisis sometimes affects the macula. In eight patients described by Yassur and colleagues three presented with foveal retinoschisis and five had macular pigmentary changes. Six patients had a normal single electroretinogram; the remainder had anormal a wave and a reduced b wave that returned to above the baseline levels.

Hereditary retinoschisis without foveal retino-

**Fig. 5** The left peripheral fundus of the proband's father discloses dendritic filariform structures of the inner retina. The macula appears intact, and foveal retinoschisis is absent.

**CASE 2**

The 35-year-old father of the proband was examined on 27 May 1987. His best corrected visual acuity was 20/20 in both eyes. Colour differentiation assessed with Ishihara plates, intraocular pressure measured by Goldmann tonometry, and anterior segments examined by slit-lamp biomicroscopy appeared normal in both eyes. Each fundus demonstrated peripheral retinoschisis and dendritic filariform structures in the inner layer of the schisis. No foveal retinoschisis was observed (Fig. 5). Visual field examination by Goldmann perimetry showed absolute depressions in the peripheral fields in both eyes.

A scotopic single-flash electroretinogram, recorded after 20 minutes of dark adaptation,
Autosomal juvenile retinoschisis is diagnostic for autosomal juvenile retinoschisis. Cibis described a family in which female monozygotic twins and their grandmother had peripheral retinoschisis without foveal retinoschisis. Their disorder was considered an autosomal form of retinoschisis with recessive or possibly even dominant transmission of inheritance. The father and daughter described here have retinoschisis without foveal retinoschisis. All members of the family live in a small, remote village. At least two consanguineous marriages have been recorded in the pedigree. Because the girl described in this report is also the product of a consanguineous marriage, the most likely mode of transmission in this family is autosomal recessive. The absence of foveal retinoschisis and the electroretinographic pattern displayed by the father and daughter also differentiate this condition from other forms of vitreoretinal dystrophy.

References


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K. Yamaguchi and S. Hara

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