X-LINKED HEREDITARY RETINOSCHISIS*†

BY

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AN X-linked degenerative retino-choroidal disease, hereditary retinoschisis, has been identified in the last 10 years. In milder cases the disease takes the form of radial macular degeneration, but in severe cases a grey sheet-like veil is attached to the detachment in the retinal vasculature, especially in the lower temporal quadrant. In differential diagnosis, according to Ricci (1960), a dominant hereditary hyaloid-retinal degeneration (Wagner, 1938) and a recessive hereditary hyaloid-tapeto-retinal degeneration (Favre, 1958) should be borne in mind.

Genealogical tables covering several generations have been described by Sorsby, Klein, Gann, and Siggins (1951), Levy (1952), Jager (1953), Balian and Falls (1960), and Gieser and Falls (1961). The last described a 13-year-old girl—possibly a latent carrier—with cystic (perhaps traumatic) macular degeneration. Rieger (1941) described a girl in whom the pathological picture was reminiscent of retinoschisis, but gave no genealogical or genetic details.

In South-West Finland, a family has been found (Fig. 1, opposite), which shows a progressive disease from childhood, with reduced vision, slight hemeralopia, restricted visual fields, and a similar condition of the fundus.

Cases Examined

PROPOSITA

V.9, a woman aged 44, had always had poor sight which had deteriorated gradually since infancy. The visual acuity in the right eye was 0.35, with + 3 D sph., - 1.5 D cyl., axis 110°. There was opacity of the posterior lens capsule (Fig. 2, overleaf).

The left eye showed a slight Y-shaped opacity of the posterior lens capsule, and the fundus appearance was similar. The visual acuity was 0.15 with + 3.5 D sph.

HER SONS

VI.21. The visual acuity in the right eye was 0.35, with - 0.75 D sph., + 1.75 D cyl., axis 0°. In the lower part of the fundus, nasally and temporally, was a thin veil showing individual retinal vessels and large ruptures.

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Fig. 1.—Genealogical table showing the family of a woman (V.9) with X-linked recessive hereditary retinoschisis. The possibility of a mild form of phenotypic manifestation of the tendency in heterozygous form—"genetic carriers"—in probands IV.5, IV.6, and V.7 cannot be excluded. The non-cystic macular degeneration of these persons may, however, be attributable to senility.

FIG. 2.—Right fundus of a woman aged 44 with hereditary retinoschisis (V.9 in Fig. 1). In the lateral upper part of the papilla is a drusen of the optic nerve. Below the papilla rises a half-transparent veil, part containing retinal blood vessels, connected with the ora serrata by strings containing blood vessels. The colour of the fundus is spotted, especially in the detached area. Some white strings run in the superficial choroidal layers or the deeper retinal layers.

The left eye showed opacity of the posterior lens capsule. A drusen was visible in the fundal papilla, and a veil containing retinal vessels was floating in the vitreous. Retinal vessels were also visible in their normal site on the retina behind the veil. There was marked choroidal sclerosis.

VI.22. The visual acuity was 0.4 in the right eye and 0.25 in the left. The maculae showed radial cystic degeneration. The retinae were otherwise thin and showed superficial, finely granular pigmentation.

VI.23. The visual acuity in the right eye was 0.1, with +4 D sph. A subcapsular opacity was seen posterior to the lens. The fundal papilla was swollen by large drusens, and two other large drusens were seen outside the papilla, one of them at a distance of 3 papilla diameters. There were many white streaks along the blood vessels, and pronounced choroidal sclerosis with granular pigmentations and white shiny spots and lines.

The visual acuity in the left eye was 1/8, with +4 D sph. The fundus similar to that of the right eye. Temporally, the retina formed a veil with large gaps.

HER NEPHEW

VI.16. The visual acuity in each eye was 1.0, with +0.75 D sph. In the lower temporal quadrant of the left eye, a triangular veil arose from the retina into the vitreous.

HER SISTER

V.7. The visual acuity in each eye was 1.0, with +0.25 D. sph. The macula of the left eye was slightly spotted.

HER AUNTS

IV.5. Vision was good. There was slight macular degeneration of the dry senile type.

IV.6. Vision was not tested. There was slight macular degeneration of the senile type.

HER FATHER

IV.3. This patient's sight had always been poor. The right eye had become blind 10 years before, and the visual acuity in the left eye had been impaired for 7 to 8 years. In the right eye the tension was 19 mm. Hg (Schiötz). There were posterior synechiae, and the lens was opaque so that the fundus was not visible. Retinal detachment had been discovered in the right eye in 1953.

The visual acuity in the left eye was 0.05, with -3 D sph. The tension was 22 mm. Hg (Schiötz). There was slight central opacity of the lens. The fundal papilla and fundus were normal, except that the macula showed a yellowish centre of degeneration well-defined by circular arcs, which was two-thirds of a disc diameter in size.

HER COUSIN

V.16. The visual acuity in each eye was 1.3. The fundus was normal except for a large pigmented area in the lower temporal quadrant immediately below the blood vessels. The retina, as seen by Goldmann's three-mirror contact lens, was of varying thickness. A small optically empty area was detected between the retina and vitreous.

Up to the middle of 1961, genealogical tables covering some seventy cases of X-linked recessive retinoschisis have been reported in the literature, all of them in males. Our proband (V.9) acquired the defect in both X-chromosomes because her parents (IV.3 and 10) were consanguineous in two different ways. It has been ascertained that all her sons by two different fathers (VI.21 and 22 and VI.23) are affected, and that her sister (V.7) is a conductor, with an affected son (VI.16).

REFERENCES

- BALIAN, J. V., and FALLS, H. F. (1960). *A.M.A. Arch. Ophthalm.*, **63**, 92.
 FAVRE, M. (1958). *Ophthalmologica (Basel)*, **135**, 604.
 GIESER, E. P., and FALLS, H. F. (1961). *Amer. J. Ophthalm.*, **51**, 1193.
 JAGER, G. M. (1953). *Trans. ophthalm. Soc. U.K.*, **73**, 617.
 LEVY, J. (1952). *Brit. J. Ophthalm.*, **36**, 626.
 RICCI, A. (1960). *Ophthalmologica (Basel)*, **139**, 338.
 RIEGER, H. (1941). *Klin. Mbl. Augenheilk.*, **106**, 638.
 SORSBY, A., KLEIN, M., GANN, J. H., and SIGGINS, C. (1951). *Brit. J. Ophthalm.*, **35**, 1.
 WAGNER, H. (1938). *Klin. Mbl. Augenheilk.*, **100**, 840.