

The hereditary hyalo-retinopathies

In the confused spectrum of diseases affecting the vitreous and retina, stretching at the one end from the developmental vitreous anomalies—an extreme example of which is persistent hyperplastic primary vitreous—and at the other end to the developmental retinal disorders—an extreme example of which are the retinal dysplasias, there lies a group of conditions known as the hereditary hyalo-retinopathies. A clear distinction on clinical grounds in this group of conditions is very difficult because of the diffuseness of the clinical signs, but fortunately distinctions can be drawn on genetic grounds. Four of these conditions, though rare, have been described in the literature.

(1) **X-linked recessive juvenile idiopathic retinoschisis** (Fig. 1) has been described under a number of names such as congenital vascular veils in the vitreous and cystic retinal detachment. The condition is characterized by retinoschisis often with large holes in its inner layer, by vitreous membranes and occasional vitreous cords and posterior vitreous detachments, by field defects corresponding to the retinoschisis, by normal visual acuity (unless the macula is affected with cystic change), and by a reduced electroretinogram.

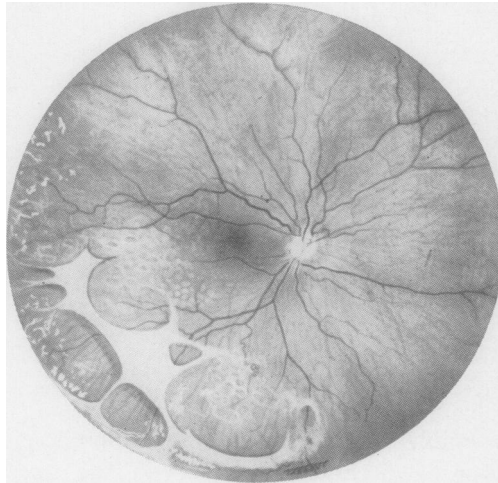


FIG. 1 *X-linked recessive juvenile idiopathic retinoschisis*

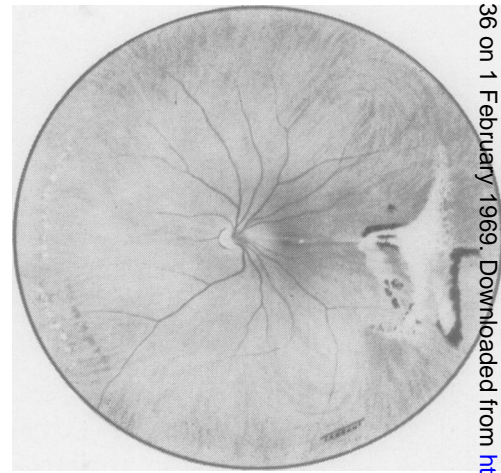


FIG. 2 *Hyalo-retinopathy of Wagner*

(2) **Autosomal recessive idiopathic retinoschisis** with night blindness is rare and is characterized by diminished visual acuity due to cystic macular change, retinoschisis with the appropriate field defects and often large holes in the inner layer of the retinoschisis, clumps of pigment in the retina resembling retinitis pigmentosa, degeneration of the vitreous with vitreous cords, an absent electroretinogram, and night blindness.

(3) **Autosomal dominant hyalo-retinopathy of Wagner** (Fig. 2) is characterized by destruction of the vitreous so as to leave an optically empty space, by some degree of vitreous detachment, and by filaments within the vitreous. Pigment clumps and pigmentation may be noticed in the retina especially along the blood vessels. Localized retinoschisis may be present especially round areas with vitreous traction, and pre-retinal white condensations have been described corresponding pathologically to glial membranes. Vision is generally good although central scotomata have been noted in the visual fields, but sometimes it is reduced by a complicated cataract. There is often some degree of myopia, the electroretinogram is sub-normal, but night vision is full.

(4) **Autosomal dominant macro-fibrillary vitreo-retinal degeneration of Favre** occurs often (although not exclusively) in young high myopes, and is associated with retinal detachment, vitreous degeneration, and some degree of cataract.

These differences are set out in the Table, but it should be noted that all four conditions tend to merge into one another. The differentiation in the vitreous pathology is seldom definite, and all may develop retinal detachment and reduced visual acuity with changes in the electroretinogram.

Table Hereditary hyalo-retinopathies

Characteristics	1	2	3 (Wagner)	4 (Favre)
Heredity	X-linked recessive	Autosomal recessive	Autosomal dominant	Autosomal dominant
Vision	Normal	Defective	Normal	Normal
Fields	Defective	Defective	Central scotomata	Normal
Macula	Rarely cystic change	Cystic change	Normal	Normal
Retina	Schisis	Schisis Retinitis pigmentosa	Peripheral schisis Pigment clumps	Detachment
Vitreous	Membranes Cords Detachment	Degeneration Strands	Destruction Glial membranes	Macro-fibrillary degeneration Myopic degeneration
Lens	Clear	Complicated cataract	Presenile cataract	Lens opacities
Refraction	Hypermetropia	—	Myopia	High myopia but may be emmetro- propic
ERG	Reduced	Absent	—	—
Night vision	Normal	Absent	Reduced	—
Key references	Gieser and Falls (1961)	Favre (1958)	Wagner (1938) Alexander and Shea (1965)	Favre (1961)

N.B.—As these conditions progress, the severer characteristics become common to all!

The difficulty in recognizing these conditions is due to three factors:

- (1) The appreciation of changes in the contour and structure of the vitreous. With the help of the slit lamp and a contact lens it should be possible to distinguish between a vitreous detachment and vitreous collapse, between vitreo-retinal adhesions and vitreous condensations either in bands or in sheets, and between lacunar atrophy and large and small fibrils.
- (2) The recognition of the manifestations of retinoschisis. The anterior leaf of the retinoschisis is very thin and diaphanous and contains retinal vessels. Often white dots may be scattered on its surface and holes which vary from quite small to very large may be present within this inner membrane. On indentation a "white with pressure" sign may sometimes be elicited. Certain retinal changes occur secondarily to the retinoschisis, such as pigment lines (which may represent the one-time border of the retinoschisis) and conglomerations of white dots. Some of these white areas may be on its surface rather than in the retina proper. Retinal oedema, local retinoschisis and a vitreous tag at the site of the conglomerated white dots may all be noted.
- (3) The appreciation of the genetic patterns of these diseases. This involves not only taking a very accurate family history so as to distinguish between the various types of mendelian inheritance (Blach and Jay, 1968), but also in examining the relatives of the patient, as the early stages of these conditions are often asymptomatic.

There is no doubt that if the above criteria are applied to new cases within this group, then further hyalo-retinopathies will be described.

References

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ILLUSTRATIONS

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