Sorsby’s pseudoinflammatory macular dystrophy

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SUMMARY Forty-three additional members of a family described originally by Sorsby and Mason have been examined in order to typify pseudoinflammatory macular dystrophy more accurately. Six new affected members were identified by history alone and a further 7 were examined. Vision is lost from disciform macular degeneration generally during the 5th decade of life. Thereafter peripheral degeneration occurs in some affected members. Before loss of vision the fundus changes include fine drusen-like deposits at the level of Bruch’s membrane, angioid streaks, and plaque-like deposits of yellow subretinal material in the macular region. These changes are different from those seen in dominant drusen.

In 1949 Sorsby and Mason1 described 5 families with dominantly inherited macular dystrophy in which affected members suffered bilateral central visual loss in the fifth decade of life. Duke-Elder and Perkins2 referred to the condition as pseudoinflammatory macular dystrophy. The maculae showed subretinal haemorrhage, accumulation of fibrous tissue under the retina, and choroidal atrophy. These changes were very suggestive of disciform degeneration, and fluorescein fundus angiography has demonstrated the presence of subretinal vascular tissue arising from the choroid.3 Sorsby and Mason1 also described progressive atrophy of the peripheral choroid and the retinal pigment epithelium in later life causing severe visual disability.

A histological study4 was performed on 2 affected members of one of the families reported by Sorsby and Mason.1 Unfortunately, conclusive evidence of dominant inheritance was not available in this family. The histological changes demonstrated were thickening of Bruch’s membrane, which was ruptured in several places, and fibrovascular invasion of the subpigment epithelial space from the choroid. Drusen were also shown in one case. Despite the clinical appearance of profound choroidal atrophy, the histological study showed atrophy of the choriocapillaris only, with relative preservation of the larger choroidal vessels. It was considered that the atrophy of the choroid was no greater than was commonly seen in the eyes of old people who showed no associated changes in Bruch’s membrane or subretinal new-vessel formation and that choroidal vascular closure was not the primary cause of the disorder.

The predisposing changes in the posterior pole have yet to be accurately defined. Younger members of these families have been reported to have pigment epithelial atrophy5 and colloid bodies.6 Deuteranomaly has also been described in young members as a dominant trait7 and this may well represent the functional correlate of these predisposing changes.

Doubt has been expressed whether the high incidence of disciform degeneration and the presence of peripheral choroidal degeneration indicate that the disease process described was fundamentally different from dominant drusen or whether these families had been selected on the basis of severity alone.8 Recently additional members of a family described in the original paper by Sorsby and Mason1 (Ewbank family) have been examined and are described in this paper.

Patients and methods

Forty-three members of one family were examined (Fig. 1): 41 were descendants of I/1 by his first marriage and the remainder by his second. Sorsby was aware of the first marriage but had no details of the descendants except that ‘there were children’.

In addition to a full clinical examination colour vision was assessed by Panel D-15, fundus were photographed, most had fluorescein angiography, and electro-oculography (EOG), electroretinography (ERG), and macular ERG9 were undertaken in

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Fig. 1  Extension of the Ewbank family pedigree, originally described by Sorsby and Mason. 

Results (See Table 1)

Six new affected members were identified by history alone: II/1 and III/1 both lost reading vision in their early 40s and had progressive loss of peripheral vision during the sixth decade of life such that they could not navigate alone by the age of 70. III/5 died at the age of 52, by which time she could not read. III/7 lost reading vision before the age of 40 but subsequently her vision remained stable. III/12 could not read after 54 years and was not independently mobile after 70 years. IV/7 had loss of RE visual acuity at 43 years followed a year later by loss of LE central vision; no further loss has occurred since.

Seven members were seen who had suffered loss of vision: IV/11 and IV/13 both lost central vision in their early 40s and thereafter had progressive loss of peripheral field. At the time of the study neither could walk unguided; each had large fibrous disciform lesions bilaterally and widespread peripheral atrophy of the retina and choroid. IV/12 lost central vision in the fifth decade of life but vision had subsequently remained stable. She had bilateral disciform lesions and densely packed fine drusen between 20-25° and from the fovea (Fig. 2). IV/11 was first seen with distortion of the left eye vision at the age of 39 years. On examination there appeared to be a confluent deposition of yellow debris beneath the macular neuroretina which was within or just deep to the retinal pigment epithelium bilaterally (Fig. 3a) and a small disciform lesion inferonasal to the left fovea. On fluorescein angiography the yellow material corresponded with choroidal hypofluorescence during the early stages of the study, becoming slightly hyperfluorescent within 10 minutes of initial dye entry (Fig. 3b-d). The major choroidal blood vessels could be seen within the lesion. The left eye showed an identical change in the appearance of the choroid and the subretinal blood vessels (Fig. 3e). The subretinal

Table 1  Summary of findings in those patients with phenotypic expression of the abnormal gene

<table>
<thead>
<tr>
<th>Patient</th>
<th>Year of birth</th>
<th>Age at visual loss</th>
<th>VA</th>
<th>Fundus changes</th>
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<tr>
<td></td>
<td></td>
<td>RE</td>
<td>LE</td>
<td>RE</td>
</tr>
<tr>
<td>IV/11</td>
<td>1907</td>
<td>43</td>
<td>41</td>
<td>PL</td>
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<td>1910</td>
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<td>1933</td>
<td>45</td>
<td>6/9</td>
<td>CF</td>
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</tr>
<tr>
<td>V/19</td>
<td>1965</td>
<td>6/6</td>
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</table>

VA = visual acuity. RE = right eye. LE = left eye.
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neovascular tissue grew despite photocoagulation with consequent loss of visual acuity (Fig. 3f,g). V/14 lost visual acuity of both eyes within a 9-month period due to disciform macular lesions. Examination showed large fibrous subretinal lesions; elsewhere in the posterior poles there were densely packed fine drusen and angioid streaks radiating from the optic discs (Fig. 4). V/16 had lost visual acuity at 38 and 39 years. She had large fibrous disciform lesions at both maculae but the fundi were otherwise normal, apart from minor retinal pigment epithelial irregularities (Fig. 5).

A further 8 subjects were seen who were at 50% risk of having the abnormal gene, had normal vision, but were under the age of 45. Of these, 3 had fundus abnormalities: V/15 and V/17 had fine drusen-like deposits at the level of Bruch's membrane, and one, V/15, had angioid streaks (Fig. 6a,b). The other (V/8), had an even yellow deposit beneath the retina.

**Fig. 2** Right fundus of IV/12 showing densely packed fine drusen around the optic disc.

**Fig. 3a**

**Fig. 3** Right eye shows an area of subtle discolouration of the fundus of the macula (a) which is hypofluorescent on fluorescein angiography with large choroidal vessels seen in the depths of the lesion (b,c). Five minutes after the injection the area is hyperfluorescent (d). Fluorescein angiogram of the left eye shows similar changes and a disciform lesion (e) which was photocoagulated but returned (f,g).

**Fig. 3b**

**Fig. 3c**
which obscured choroidal fluorescence during the early part of fluorescein angiography but not in the later stages. This change was similar to that seen in his mother (V/11). None had abnormalities of EOG light induced rise in ocular potential, mass ERG, or macular ERG. Colour vision was also normal in all as tested on the Panel D-15.

Discussion

As a result of this study several conclusions can be drawn. The pattern of visual loss is identical to that described by Sorsby and Mason\(^1\) and subsequent papers\(^2\) with loss of central vision due to disciform lesions occurring in the fifth decade of life. The earliest visual loss identified in this family was 37 years (V/16), and good visual acuity was maintained in one patient into the sixth decade of life (III/11). The interval between visual loss in the 2 eyes varied from 9 months to 10 years.

The subsequent progressive peripheral chorioretinal atrophy causing loss of peripheral field is well...
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Fig. 4 Right eye of V/13 with a large disciform lesion and angioid streaks radiating from the optic disc.

Fig. 5 V/16 showing a disciform lesion at the posterior pole, but the fundus otherwise appears relatively normal.

Ashton and Sorsby describe the significance of this atrophy and conclude that choroidal vascular closure is unlikely to be the primary event in this order. The alternative explanation is that this atrophy is second-

described by Sorsby and Mason but has received little emphasis since that time. This was identified in 5 (3 by history) out of 8 affected patients reaching the age of 70 years and caused social blindness in each.

Fig. 6a

Fig. 6 Red-free (a) and fluorescein angiogram (b) of the right eye of V/16 showing diffuse, fine drusen-like deposits and angioid streaks radiating from the optic disc.
filling is delayed
occur in drusen, from
tion of disease.
related to material suggests and in whole of Abnormal hypofluorescence. Drusen-like deposits, evenly seen outside that obscuration transit of choroidal obscuration themselves in involvement loss and visual in alternatively affected members of the disciform area. The disciform lesion in each affected member seen and the rapid loss of vision in older relatives implies a similar process. No patient has yet been recorded in whom central visual loss was due to geographic atrophy. The disciform lesions were always large, which contrasts with many other conditions causing disciform lesions in the young. This may be a reflection of diffuse rather than multifocal disease at the level of Bruch’s membrane in this disorder.

The evidence in this family suggests that pseudo-inflammatory macular dystrophy as described by Sorsby and Mason is a specific autosomal dominant disease distinct from dominant drusen and from other autosomal dominant fundus dystrophies.

This work was supported in part by MRC grant no. G796/618. A.H. was aided by the A. E. Baker grant (Canadian National Institute for the Blind).

References
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