INHERITED MACULAR CYSTS*

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THIS dominantly inherited condition presents, in its early stages, a unique ophthalmoscopic picture, and has a progressive course during which the appearance of the lesions alters considerably.

The following cases are reported because it is thought that this family study (Fig. 1) illustrates some of the cardinal features of the condition, and includes the youngest case so far recorded in the literature. The different stages of the condition seen in members of this family seem to follow the course described by Sorsby, Savory, Davey, and Fraser (1956). In the following case reports, only those features which are directly related to the macular disease will be mentioned. All these patients have brown irides, and none have defects of colour vision.

FIG. 1.—Family tree.

Case Reports

Case 1, a girl aged 8 years, the proband, presented with a right convergent squint of 15° of recent onset. The visual acuity was 6/24 in the right eye and 6/9 in the left.

At the right macula was an area of retinal disturbance surrounded by a circular "watermark" (Fig. 2, overleaf), and at the left macula a perfectly circular, raised, yellowish-coloured swelling about 2 disc diameters in width (Fig. 3, overleaf).

Fixation was eccentric in the right eye at the superior edge of the "watermark", and in the centre of the raised area in the left. The central visual fields showed a central scotoma in the right eye, the left eye being normal.

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Case 2, a girl aged 11 years, the proband's elder sister, had a visual acuity of 6/9 in the right eye and 6/18 in the left. In the right macular region was an oval area of disturbance, the lower part showing a gravitational level of yellow cyst content, the cyst having apparently burst and partly emptied (Fig. 4, opposite). The left macula showed central aggregations of retinal pigment surrounded by a "watermark" (Fig. 5, opposite).

Fixation was eccentric in both eyes at the superior edge of the "watermark". The central visual fields showed bilateral central scotomata.

Case 3, a girl aged 3 years, the proband's younger sister, had bilateral perfectly circular, raised, yellowish swellings at the macula about half a disc diameter in width (Figs 6 and 7, opposite).

Case 4, a man aged 38 years, father of cases 1, 2, and 3, had a visual acuity of 6/4 with correction in both eyes. At both maculae there were very localized minimal but definite pigmentary changes (Figs 8 and 9). The central visual fields were normal.

FIG. 8.

FIG. 9.

Case 5, a man aged 76 years, the proband's paternal grandfather, had a visual acuity of 6/60 in both eyes, and early lens changes. There was bilateral central pigmentary degeneration with choroidal sclerosis, the circumscribed nature of the lesion being still visible (Figs 10 and 11).

FIG. 10.

FIG. 11.

Case 1.

FIG. 2.

FIG. 3.

Case 2.

FIG. 4.

FIG. 5.

Case 3.

FIG. 6.

FIG. 7.

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The proband's mother, paternal uncles, and paternal grandmother were all normal. We were not able to examine the only other surviving member of the paternal family, the proband's great-aunt.

It was reported by Case 5 that his paternal grandfather was supposed to have been blind, but the cause was unknown.

Discussion

From this family and the series reported by Barkman (1961), whose youngest patient was a girl aged 2 years, we can postulate the progress of the lesion. Barkman was fortunate enough to watch the lesions develop in his case from a pinkish macular disturbance noted 4 days after birth; he describes these earliest changes as:

“minute alterations with pink transparencies in retinal pigment alternating with agglomerations of pigment within a central region of $\frac{1}{8}$ disc diameter, with a slight protrusion of the overlying retina” (Personal communication).

From this protrusion the lesion gradually developed into a yellowish appearance at 2 years of age.

The lesions increase in size without otherwise altering, probably reaching a maximum of 2 disc diameters in most cases, generally between 8 and 10 years of age, although this stage may vary considerably and may develop as late as 43 years (Sorsby and others, 1956).

The cystic appearance then suddenly changes. According to Sorsby and others (1956), the cysts burst leaving an area of pigmentary disturbance surrounded by a circular “watermark” at the limits of the cyst; this persists throughout life, though non-specific degenerative changes may become superimposed upon it later. Fig. 4 (Case 2) shows a cyst that seems to have burst but has become only partly emptied of its yolk-like contents.

Our series demonstrates that this affection is inherited as an autosomal dominant with high penetrance, so confirming the view of Sorsby and others (1956) and agreeing with that of the majority of authors on the mode of inheritance of macular anomalies of this type. The case examined 4 days after birth by Barkman (1962) would appear to confirm fairly conclusively the congenital nature of the affection, as does that of our youngest patient aged 3 years (Case 3). It seems probable that Case 4 represents a carrier state, or is an example of a variation in the expression of the faulty gene.

Klien (1950) described an ophthalmoscopically similar lesion as an extensive defect of the first neurone. This is unlikely to apply to our cases, as the visual acuity remains relatively little altered while the cyst is intact. It seems certain that the lesion does not involve the neuro-epithelium in the early stages. In Fig. 3 (Case 1) the left fovea can be seen dimpling the large yolk-like cyst. This eye was hypermetropic (+5 D) and the visual acuity was 6/9, whereas that of the less hypermetropic (+3.5 D) right eye, in which the cyst had ruptured, was 6/24.

Renard, Dhermy, and Amar (1960), describing an ophthalmoscopically similar lesion, were convinced after slit-lamp examination that the lesion was flat, but we were in no doubt that the lesions in our cases were raised.

Visual acuity appears to be only minimally affected while the cysts are intact, but drops sharply when the cystic formation disappears, and this drop is accompanied by eccentric fixation, usually above the area of the lesion as in Case 1 (right eye) and in Case 2 (right and left eyes).

Conclusions

Our five cases, occurring in three generations, show four types of morphology:

- (1) Central cystic
 - (2) Central atrophic
 - (3) Central pigmentary: Case 4
 - (4) Central retinal and choroidal: Case 5
- } Cases 1, 2, 3

It would be possible to see each of these lesions in unrelated individuals and classify them morphologically as though they were different entities, and this has generally been done in the past; *e.g.* Case 3 (Figs 6 and 7) would be called inherited cystic macular degeneration (Sorsby and others, 1956), Case 1, right eye (Fig. 2) and Case 2, right and left eyes (Figs 4 and 5) could have been classified as Stargardt's atrophic macular dystrophy, and Case 5 (Figs 10 and 11) could be classified as senile central choroido-retinal atrophy.

It would seem, therefore, that this series lends weight to the contention of many present-day workers, *e.g.* François (1961) and Barkman (1961), that many types of central retinal degeneration are of an inherited nature and comprise a single nosological entity although varying greatly in ophthalmoscopic appearance.

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