FAMILIAL MACULAR DEFECTS*

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THE description of individual disorders of the macula by various names has led to confusion. Several familial and hereditary macular diseases have been classified under the title of heredo-macular degeneration. In this category is Best's disease, which is described as a familial congenital macular defect. This is a rare condition, characterized by retention of good central vision and lack of progression of the lesions.

Varying degrees of abnormality of the macula have been noted in different members of the same family, the clinical appearances resembling those described in Best's disease (congenital macular and paramacular atrophic defect).

Case Reports

Case 1, a boy aged 15 years, the first member of the family to be observed, was first seen in August, 1957.

Records show that when the patient was examined in January, 1948, at the age of 6, he was found to have a left convergent squint and an unaided visual acuity of 6/36 in the right eye and 6/60 in the left. Retinoscopy under atropine cycloplegia revealed, in both eyes, hypermetropia of the order of 11 dioptres, and "coloboma" at the macula. In February, 1955, when he was aged 13, the visual acuity with glasses was recorded as 6/18, 6/12 partly in the right eye and 6/60, 6/36 partly in the left.

When the patient was seen at Moorfields in August, 1957, the visual acuity in the right eye with $+8.0/+1.0$ 90° was 6/36, and in the left eye, with $+7.5/+1.0$ 90°, 6/24 partly. The ocular movements were normal, and the media clear. The right macular region showed a central atrophic area surrounded by a ring of pigment, which in turn was bordered by a band of atrophic retina (Fig. 1). In the left eye, a circular atrophic area, slightly larger than the disc, was apparent at the macular region (Fig. 2).

FIG. 1.—Case 1, right eye.

FIG. 2.—Case 1, left eye.

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Examination with the Hruby lens confirmed the ophthalmoscopic impression of an atrophic lesion.

Investigations carried out at the Institute of Ophthalmology in February, 1957, failed to reveal any evidence of systemic disease or infection which could be responsible for, or associated with, the eye condition.

Case 2, a man aged 47 years, the father of Case 1, was found to have poor sight. He was employed on manual work, which was unexact from the ocular standpoint, and could give no precise information as to the duration of his disability. In 1948, Mr. Gordon noted that the maculae of this patient showed a clinical picture similar to that seen in his son, and the visual acuity was then 6/60 in the right eye, and 6/36 in the left. The patient stated that, of recent years, the sight in the right eye had deteriorated. On examination the right eye was divergent, and the visual acuity was hand movements only. Fundus examination showed an extensive flat detachment inferiorly, with a typical "hole" at the macula. The left eye had an acuity of 6/36 partly, which was not improved by glasses. Ophthalmoscopically, a small ring of pigment with some irregular pigmentation surrounding it, was evident in the macular area (Fig. 3).

FIG. 3.—Case 2, left eye.

FIG. 4.—Case 4, left eye.

Case 3, a girl aged 18 years, the elder sister of Case 1, was examined and found to have normal eyes.

Case 4, a girl aged 12 years, a younger sister of Case 1, had eyes which were externally normal, but a marked esophoria was elicited on cover test. In the right eye, the visual acuity with correction was 6/9, and in the left eye 6/36. Fundus examination revealed spherical cystic swellings at both macular regions (see Fig. 4 and Fig. 5, opposite). The oedema was visible with the Hruby lens.

Case 5, a girl aged 9 years, a younger sister of Case 1, showed a constant left convergent strabismus. The visual acuity in the right eye with correction was 6/6 partly. The media were clear, but the macula showed a slight swelling, much less than in Case 4. Unfortunately a fundus photograph of this eye could not be obtained. The visual acuity in the left eye was reduced to "counting fingers" at one foot. The fundus showed a clear-cut circular atrophic area at the macula, similar to that in the left eye of Case 1.

Discussion

The clinical appearance of the eye lesions in certain members of this family bear a close resemblance to those found in Best's disease, which is

described as a congenital macular and paramacular atrophic defect. According to Sorsby (1951), the eye lesions in Best's disease are stationary, frequently with good central vision.

FIG. 5.—Case 4, right eye.

In this family, the eyes showing atrophic defects have poor central vision. The appearances in Case 4 are exudative rather than atrophic, and may represent an earlier stage of the atrophic lesions, in which event the condition is not a congenital one. In Case 5, the appearances in one eye strongly suggest an early exudative lesion, while an atrophic lesion is present in the other. The clinical picture in the right eye of Case 1 and the left eye of Case 2 is consistent with the pigmentary reaction following the bursting of a retinal cyst. This supposition is strengthened by the finding of a macular "hole" in the right eye of Case 2, and suggests that some macular "holes" are in fact variants of Best's disease.

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REFERENCE

SORSBY, A. (1951). "Genetics in Ophthalmology". Butterworth, London.