FUNDUS FLAVIMACULATUS*† TWO FAMILIAL CASES WITH MACULAR DEGENERATION

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Two cases of a familial fundus lesion in sisters are reported. The fundi show scattered pale flecked lesions and exudation at the maculae. These were observed for several months and investigated by fluorescence retinal photography.

Case Reports

Case 1, a married woman aged 46, the elder of two sisters, was the first to be seen. She complained of sudden blurring and distortion of the vision of the left eye; the vision in the right eye had been poor since she had had a corneal ulcer at the age of 13 years. Her general health was good. Her mother's vision had become poor in middle age and was very poor at the time of her death. She had three brothers and one sister all with normal vision. The sister, however, subsequently attended with similar symptoms.

Examination.—In November, 1965, the visual acuity was 6/60 in the right eye improving to 6/18 with a pinhole, and 6/18 in the left eye with no improvement. The right cornea showed deep stromal scarring. The left fundus had a subretinal haemorrhage about two disc diameters in size situated immediately above the macula. Both fundi showed a diffuse pleomorphic flecked appearance which is fully described below in conjunction with colour and fluorescence retinal photography.

Laboratory Investigations.—Hb 106 per cent., white blood cells 7,000 with a normal differential count, Wassermann reaction and Kahn test negative, serum cholesterol 190 mg. per cent.

The visual acuity in the left eye continued to deteriorate and 2 months later was reduced to counting fingers. At this time the haemorrhage at the left macula had disappeared leaving an appearance of disciform degeneration of the macula. 11 months after the first examination, the disciform lesion showed microaneurysms at its margin and was surrounded by a ring of hard exudates in a circinate pattern. The right fundus had not changed. Colour and fluorescence photography were carried out at this stage; these are described below.

Case 2, a married woman aged 43, the younger of the two sisters, attended 3 months later. She complained of sudden blurred vision in the right eye 3 weeks previously. Her general health was good.

Examination.—In February, 1966, the visual acuity was 6/24 in the right eye, not improved with a pinhole, and 6/24 in the left, improving to 6/12 with a -1 D sph., -1.75 D cyl., axis 180°. The right fundus showed subretinal haemorrhages above and below the macula which was oedematous. Both fundi showed the pleomorphic flecked appearance described in the sister.

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Laboratory Investigations.—Hb 104 per cent., white blood cells 5,000 with a normal differential count, Wassermann reaction and Kahn test negative, serum cholesterol 210 mg. per cent. The left central and peripheral fields were normal. The right showed a central scotoma with a full peripheral field.

The visual acuity in the right eye progressively deteriorated to 6/60 in the next 4 months, the left eye remaining unchanged. At this time the right fundus showed the remains of the haemorrhage and persisting oedema at the macula. After 7 months the visual acuity in the right eye had further declined to hand movements. At this stage, when fluorescence retinal photography was performed, the macula had taken on the disciform appearance as seen in the left eye of Case 1, but was not surrounded by a ring of hard exudates.

Colour and Fluorescence Retinal Photography

This was carried out in both patients, and as the lesions are essentially similar the results in the two cases will be described together. The fundus lesions consist of four components: deep flecking, colloid deposits, atrophic areas, and exudative macular lesions; the last affected the right macula in Case 2 and the left in Case 1. Fig. 1 illustrates the combination of flecking with colloid deposits at the macula in the less affected eye of Case 2. Fig. 2 illustrates a more dispersed exudative lesion at the macula of the left eye of Case 1 surrounded by an incomplete ring of hard exudate.

Fluorescence retinal photography was confined to the macular areas of both eyes in the two patients, and the descriptions relate only to the macular lesions. The areas of flecked retina showed a mottled background fluorescence which faded with the transit of dye, no residual fluorescence was seen in the late pictures of the less affected eye of Case 2 and only one small mottled area in that of Case 1. In the areas included in the photographs there was no evidence of fluorescence of the colloid spots. The areas of atrophy were not especially sought in the fluorescence photographs, but one small area above and lateral to the left macula of Case 1 showed relative background pallor and the presence of fluorescent choroidal vessels (Fig. 3). The worse affected maculae of the two patients showed slightly differing appearances which are seen in the early phase of transit in Fig. 3 (Cases 2 and 1 respectively). In Case 2 a moderate area was shielded from choroidal fluorescence which began to leak profusely at an early stage of the transit. In case 1 a much larger area was shielded from fluorescence with some particularly dense fluorescent spots scattered irregularly around its periphery. Later, slow leakage occurred in this area and filled it irregularly.

Discussion

The term fundus flavimaculatus was first used by Franceschetti (1962); the original description was followed by many papers and the subject was well reviewed with a report of 27 cases and one histological examination by Klien and Krill (1967). Although there was at first a tendency to consider that the fundus lesions described in this condition were similar in character to colloid deposits (Krill and Klien, 1965) and could be included with other conditions such as Doyme's honeycomb choroiditis, malattia Levantine, Tay-Hutchinson choroiditis, etc., the establishment by Klien and Krill of a distinct histological pattern, with the presence of mucopolysaccharide deposits in the inner layers of the retinal pigment epithelium, has added weight to the clinical distinction now being made on the basis of morphology and fluorescent appearance. Morphologically, fundus flavimaculatus presents indistinct "fishtail or pisciform" deposits which later fuse and become less marked,

FIG. 1.—Reproduction of a colour transparency of the left macula in Case 2. The principal feature is a widespread mottled deposit with soft edges, fancifully described as "pisciform or fish-tail". The macula appears relatively free of deposits but there are sharply outlined small colloid spots around it.

FIG. 2.—Reproduction of a colour transparency of the left disc and macula in Case 1. A large exudative lesion with an incomplete ring of circinate hard exudates occupies most of the right-hand half of the illustration; elsewhere a fine soft mottled deposit can be seen.

FIG. 3.—Fluorescence retinal photograph taken during the arterial phase after the first injection of fluorescein, left macula of Case 1. An extensive area of shielding of fluorescence occupies almost the entire photograph; marginally a flecked background pattern can be seen. Just above and to the right of the macula, which is situated centrally, an area of atrophy allows underlying choroidal vessels to be seen. Intensely fluorescent spots are seen around the periphery of the area of shielding, and below to the left early leakage is occurring in association with some of the spots; this later intensified and spread irregularly to the rest of the shielded area.

in contradistinction to the round colloid deposits; the latter retain their individual form even on coalescence. Fluorescence retinal photography in fundus flavimaculatus reveals widespread background mottling. Colloid deposits show a punctate pattern which does not correspond exactly to the visible lesions, as there is usually an excess of fluorescent spots, but occasional deposits remain without fluorescence.

The two cases reported fit the described pattern closely and belong to the more severe, and common, sub-group with associated macular disease. The presence of atrophic patches in the fundus was particularly noticed by Carr (1965) and has received little attention from other authors. The genetic status of the disease is uncertain, autosomal dominant and recessive patterns being suggested. The present records are insufficient to contribute to this knowledge, as it was not possible to examine the other members of the family.

Summary

(1) Two cases of fundus flavimaculatus in sisters are reported with photographic and fluorescence records.

(2) The disease having recently been reviewed in the literature, the two cases are discussed in the light of present opinion.

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