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Butterfly-shaped pigment dystrophy of the fovea associated with subretinal neovascularization

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Abstract ● **Background:** The association of butterfly-shaped pigment dystrophy of the fovea, an uncommon inherited macular disease, with subretinal neovascularization has rarely been reported in the literature. ● **Case report:** We describe the clinical history of a patient affected with butterfly-shaped pigment dystrophy of the fovea, myopia, and optic nerve head dysplasia. She was followed up for 23 years. During the course of the disease, bilateral subretinal neovascularization in the macular area occurred. Fluorescein angiography

confirmed the diagnosis. Recently, indocyanine green (CG) videoangiography was also performed. Because of the bilateral subfoveal localization no laser treatment was advised. ● **Discussion:** Usually, good visual acuity is maintained in this uncommon inherited macular disease. However, acute visual loss can be caused by the ingrowth of subretinal new vessels. Therefore, if visual acuity decreases or metamorphopsia develops in these patients, careful biomicroscopic examination and fluorescein/ICG angiography is advisable.

Introduction

“Butterfly-shaped pigment dystrophy of the fovea” was described for the first time by Deutman et al. [11] in 1970. The characteristic symmetric bilateral pigmentation in the deeper layers of the central retina, a markedly subnormal EOG, normal or only very slightly diminished visual acuity, and an apparent autosomal dominant inheritance pattern are fundamental for the diagnosis [11]. Because of their similar clinical appearance, butterfly-shaped dystrophy [11, 16, 20], reticular dystrophy [7, 10, 18, 24], macroreticular dystrophy [17], fundus pulverulentus [25], and adult-onset foveomacular vitelliform dystrophy [12] could be grouped together in a single pathologic entity: pattern dystrophy of the retinal pigment epithelium [27]. The concomitant presence of morphologically different pattern dystrophies either in the same affected family [13, 14, 16, 19] or in various stages of the disease in a single patient [6] suggests that they might be different expressions of the same disorder. Histopathologic and biochemical studies will probably

demonstrate these clinical impressions. A deletion in the RDS (retinal degeneration slow) gene in patients affected with butterfly-shaped dystrophy has already been reported [21].

Although the presence of subretinal neovascular membrane in patients labeled with a generic diagnosis of pattern dystrophy has been already reported [15, 23, 26], only Burgess [4] described two cases where a well-defined butterfly distribution of yellowish material in the pigment epithelium was present. In our report we describe the clinical history of a young patient affected with a dark butterfly-shaped dystrophy of the fovea and dysplasia of the right optic nerve head who developed a bilateral subfoveal neovascular membrane.

Case report

A 4-year-old girl was seen for the first time in November 1972. The ophthalmic examination revealed a positive angle kappa, simulating divergent strabismus, with good binocular vision and congenital horizontal nystagmus. The corrected (−1 sph) visual acuity

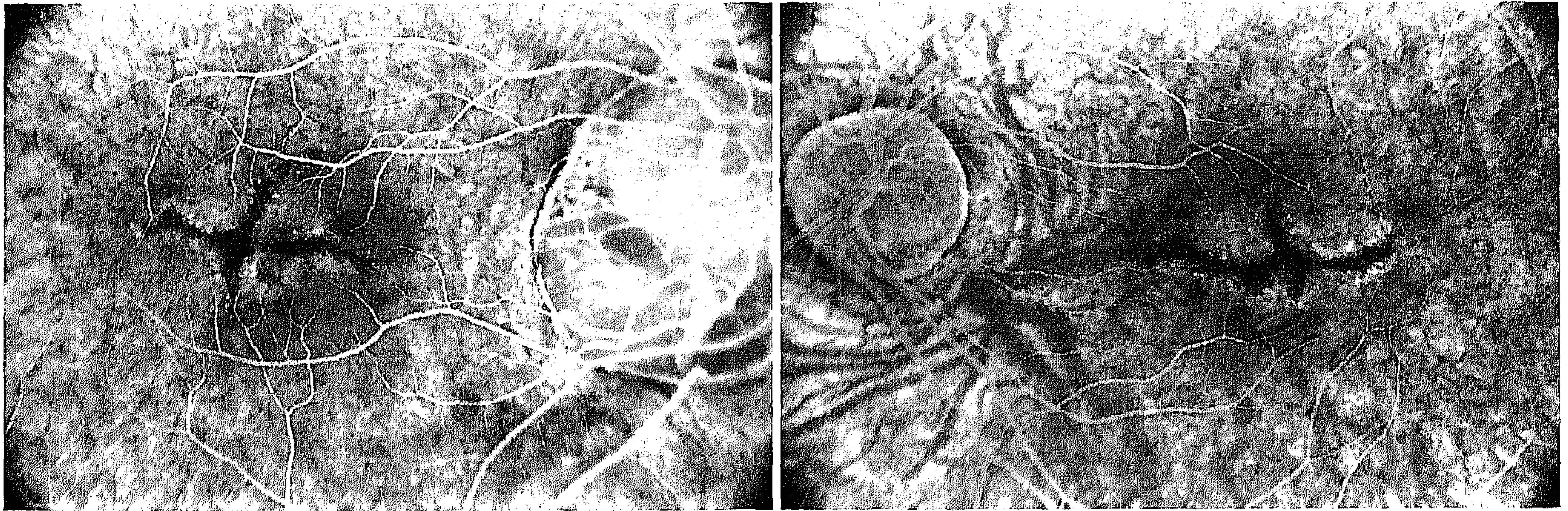


Fig. 1 *Left and right:* The early phases of high-magnification fluorescein angiography show bilateral, symmetrical, dark, butterfly-shaped pigment dystrophy of the fovea. *Left:* Evident dysplastic optic nerve head with a large inferior staphyloma in the right eye

was 0.6 in both eyes. The media were clear, and bilateral mydriatic isocoric pupil with normal reaction was reported. Ophthalmoscopy revealed a slighter pigmented fundus and a butterfly-shaped pattern of hyperpigmentation in both macular areas. Bilateral dysplastic optic nerve head with a large inferior staphyloma in the right eye was noted. The diagnosis of butterfly-shaped pigment dystrophy of the fovea was confirmed by an abnormal electro-oculogram EOG; light peak/dark ratio 128 in right eye and 163 in left eye) and a normal electroretinogram. The general medical history was unremarkable. The patient's parents and sister did not show any fundus abnormalities.

The girl was then examined in April 1982, July 1984, and November 1986. At the last visit the corrected (right eye -6.25 sph $= -0.75$ cyl $\times 170^\circ$, left eye -6.25 sph $= -0.75$ cyl $\times 165^\circ$) visual acuity was 0.8^- in the right eye and 0.6^{++} in the left eye. High-magnification fluorescein angiography [1] was then performed (Fig. 1).



Fig. 2 *Left:* Arterial phase of conventional fluorescein angiography shows a hyperfluorescent zone in the macular area of the left eye due to a circular subfoveal neovascular net. *Right:* Late hyperfluorescence caused by leakage from the choroidal neovascularization under the sensory retina

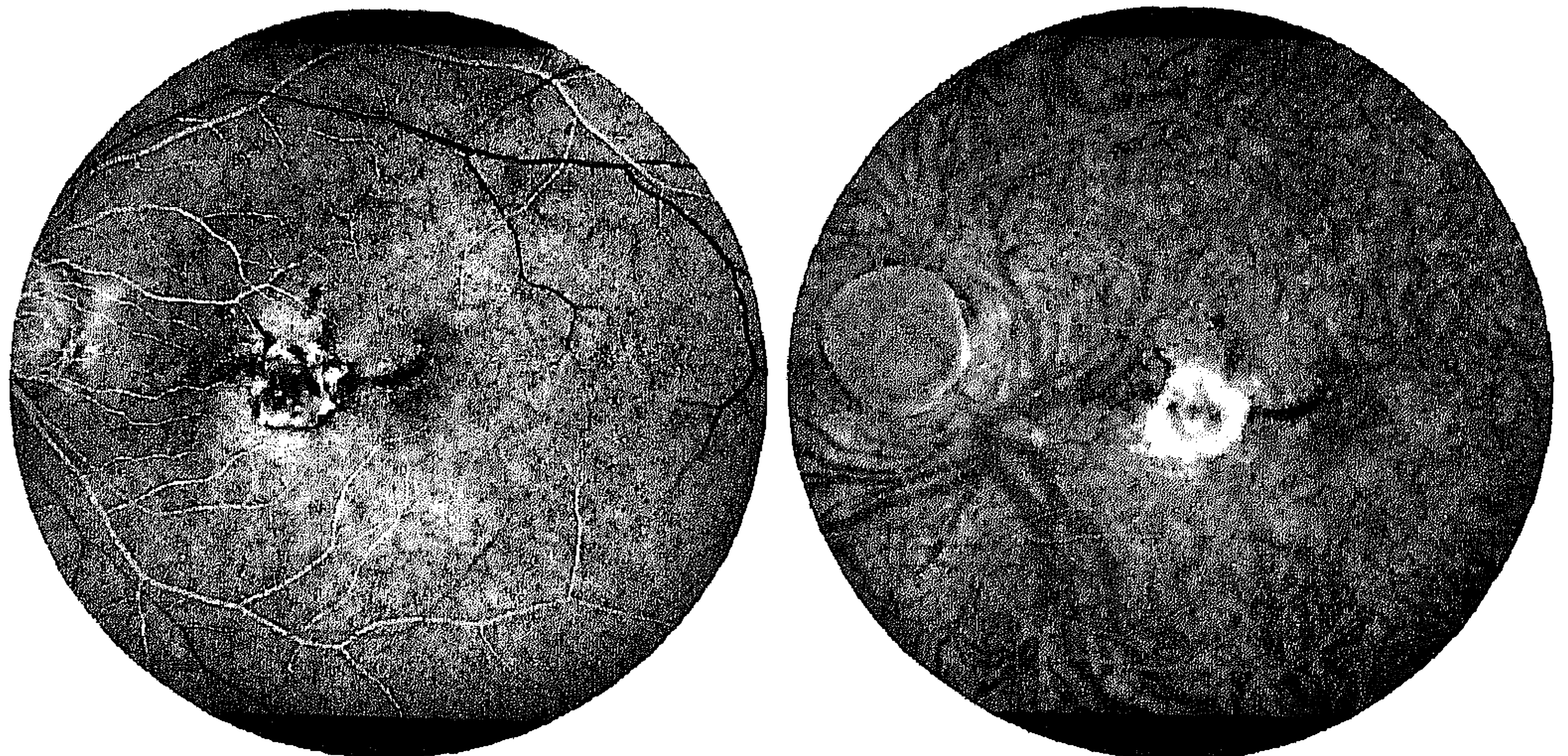
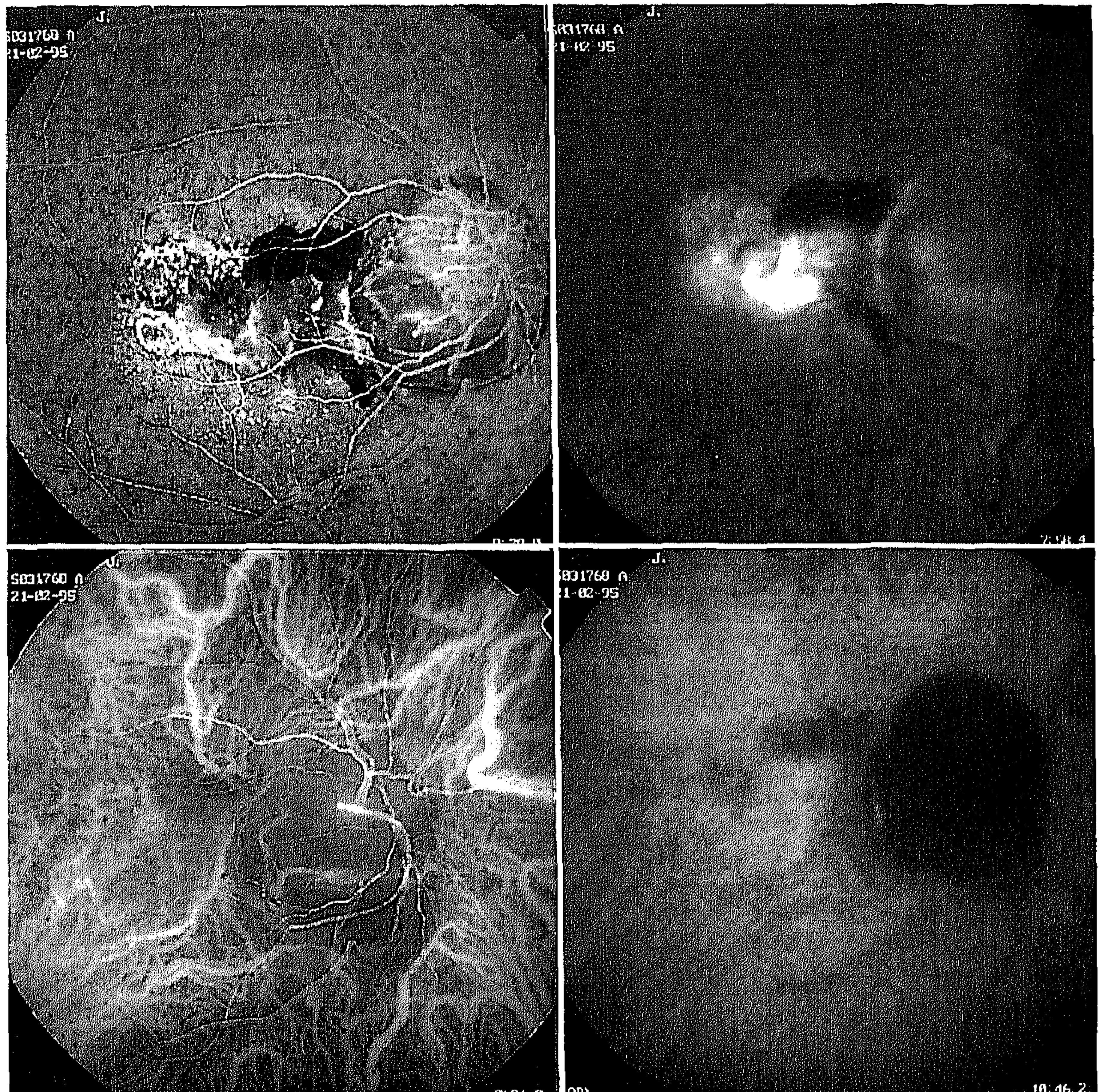


Fig. 3 Despite the lower visual acuity in the right eye, only a slight enlargement of the hyperpigmented butterfly-shaped lesion was noted in comparison to the previous angiography (Fig. 1 *left*)

Fig. 4 *Top left:* Early phase of digital fluorescein angiography shows a hyperfluorescent pattern in the macular area due to an irregular neovascular net partially obscured by a subretinal hemorrhage. *Top right:* Late hyperfluorescence caused by leakage from the choroidal new vessels with staining of the dye in the subretinal space. *Bottom left:* ICG videoangiography, penetrating the thin subretinal hemorrhage, demonstrates the choroidal layer which is partially indistinct beneath the sensory retinal detachment caused by the neovascular net. *Bottom right:* ICG videoangiography at 10 min shows a hyperfluorescent spot consistent with the more active part of the neovascular membrane



In September 1989 the corrected visual acuity was 0.3 in the right eye and 0.02 in the left eye with metamorphopsia and excentric fixation. Examination of the left fundus showed a localized macular detachment of the sensory retina with suspected subfoveal choroidal neovascularization. On conventional fluorescein angiography a circular subfoveal neovascular net was evident (Fig. 2). Because of the subfoveal localization no laser treatment was advised. Despite the lower visual acuity, only slight changes were noted in the right fundus (Fig. 3).

In September 1993, the patient experienced acute visual loss in her right eye. The best corrected visual acuity was 0.2 in the right eye, with metamorphopsia and central scotoma, and 0.02 in the left eye. Fundus examination revealed a macular neuroretinal detachment caused by a choroidal neovascularization associated with subretinal hemorrhage in the right eye. A circular, one-third disk diameter central fibrous scar was present in the left eye. No fluorescein angiography is available. Because of the subfoveal localization, no laser treatment was suggested.

Because of a sudden further visual loss in the right eye, digital fluorescein and indocyanine green (ICG) videoangiography (Topcon IMAGENet H1024 Digital Imaging System) was performed in February 1995. The butterfly-shaped hyperpigmentation was barely visible, and an irregular atrophic area with mottled pigment epithelium in both macular areas was found. The early and late phases of digital fluorescein angiography were consistent with an irregular subfoveal neovascular membrane partially obscured by subretinal blood in the right eye. This finding can be considered as an acute reactivation of the previous neovascular membrane observed in 1993. A circular fibrous central scar was seen in the left eye. Digital ICG videoangiography in the late phases clearly showed a hyperfluorescent area consistent with the more active

part of the choroidal neovascularization in the right eye (Fig. 4), while in the left eye no hyperfluorescence was displayed. At that time the corrected visual acuity was 0.16 in the right eye and unchanged in the left eye.

Discussion

In 1981 Burgess [4] described two unrelated patients with butterfly-shaped distribution of yellow material deep to the retina and an abnormal EOG with secondary ingrowth of subretinal neovascular membranes and marked visual loss in the affected eyes. Recently, Shiono et al. [23] reported a family in which one member had reticular hypofluorescent lesions extending in the macular area and nasal to the disc in both eyes, associated with bilateral macular subretinal neovascularization. Two or other members of the family showed an abnormal EOG, but no fundus abnormalities.

The histopathologic study [15] of two surgically excised subretinal neovascular membranes of patients affected with not well specified pattern dystrophy showed that membranes from patients with myopia and pattern dystrophy are composed of similar constituents. Also, Thomas et al. [26], in a report about the visual results

after surgical removal of subfoveal choroidal neovascular membranes, included two eyes affected with pattern dystrophy. In neither study was further information presented. In our case we did not think that surgical removal of the subretinal neovascularization would benefit the patient.

In our case the uncommon association of butterfly-shaped pigment dystrophy of the fovea, myopia, optic nerve head dysplasia, and bilateral subretinal neovascularization is present. It is not clear how the myopic changes influenced the development of the subretinal neovascularization. Except for the inferior staphyloma in the right eye and a slight annular peripapillary chorioretinal atrophy in the left eye, neither lacquer cracks nor chorioretinal atrophic areas were present. Notwithstanding this, delayed choroidal blood flow and choriocapillaris abnormalities typical of pathologic myopia [2, 8, 22], associated with the macular dystrophic retinal pigment epithelium (RPE) changes in both eyes of our patient, might stimulate the ingrowth of new vessels. Furthermore, whereas the neovascular net in the left eye could be consistent with myopic choroidal neovascularization, the relatively large and irregular neovascular membrane that developed in the right eye does not have the appearance of a typical myopic feature.

Several authors have described peripapillary subretinal neovascularization associated with true coloboma of the optic nerve [3, 9, 28]. In our case the subretinal net was totally separated from the border of the inferior dysplastic optic nerve staphyloma in the right eye. Therefore, we believe that these are unrelated phenomena.

In all the patients affected with pattern dystrophy, the RPE seems to be greatly compromised, but no histopathologic studies are available to exactly disclose the morphologic alterations of the dystrophic RPE. The changes observed in an experimental rat model for retinitis pigmentosa suggest a progressive breakdown of the pigment epithelial cell junctions [5]. The same pathologic course might be proposed for these different but somewhat related pattern dystrophies of the RPE, which usually show relatively slow progression. In these disorders good visual acuity is often maintained, but acute visual loss can be caused by the ingrowth of subretinal neovascularization. Therefore, informing these patients about the symptoms related to the development of macular new vessels is advisable. If visual acuity decreases or metamorphopsia develops, biomicroscopic examination and fluorescein/ICG angiography can clarify the diagnosis.

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