Principles and Practice of Clinical Electrophysiology of Vision

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Pattern Dystrophies

Michael F. Marmor

The term *pattern dystrophy* was suggested by the author[13] and Hsieh et al. in 1977[9] to describe a group of related dystrophies of the retinal pigment epithelium (RPE) that are characterized by granular or reticular pigmentation patterns and a benign clinical course. Earlier literature had described a variety of RPE dystrophies with unusual pigmentary patterns such as reticular dystrophy,[16] butterfly dystrophy,[4] fundus pulverulentus,[17] dystrophia macroreticularis,[14] and others. Although these entities may well represent different genetic disorders (pedigrees with both dominant and recessive inheritance have been described), they behave in a similar fashion with respect to RPE involvement and limited functional changes.[11] The designation "pattern dystrophy" provides a way of describing and categorizing this otherwise disparate group of disorders until specific biochemical or genetic markers become available.

CLINICAL FINDINGS

The pattern dystrophies represent inherited disorders in which there is a primary granular or reticular disturbance of the RPE without any predisposing factor such as age-related macular degeneration, pigment epithelial detachment, vitelliform macular lesions, juvenile macular dystrophy, or other secondary causes of pigmentary dispersion in the fundus. The pigmentary patterns can vary widely, and some families have extensive peripheral changes as in reticular dystrophy[16] (Fig 92–1), whereas in other families the pigmentation is limited to the macula (Fig 92–2). Families have been described in which there are individual differences among the members in their pattern of pigmentation[1, 3, 9, 13] (Fig 92–3).

Reports have also appeared of families in which a pattern dystrophy appeared to coexist with vitelliform dystrophy.[5, 7, 8] This author is rather skeptical of the proposal that these disorders fall on a continuum since the vast majority of cases in each category do not overlap and the EOG is nearly flat in vitelliform dystrophy (regardless of the extent or lack of fundus changes) while it is only somewhat reduced in pattern dystrophies (even when the pigment pattern is extensive). Some of the cases in question may represent individuals in whom the "scrambling" of a yolk has left a distinctive pigmentary pattern or individuals with an unusually severe variant of pattern dystrophy in which the macular RPE damage evolves into a pseudohypopyon (as may occur with severe drusen[6, 12]).

The pattern dystrophies typically cause few symptoms during youth or early adult years.[1, 3, 10, 11, 19] Visual acuity may be mildly subnormal, but severe visual loss as often characterizes pigment epithelial detachment, Stargardt's disease, or vitelliform dystrophy would be unusual and would in general argue against the diagnosis. Some older individuals have developed macular degenerative changes[3, 13, 19] and significant visual loss (see Fig 92–3), but others have not,[3, 19] so it is unclear whether pattern dystro-

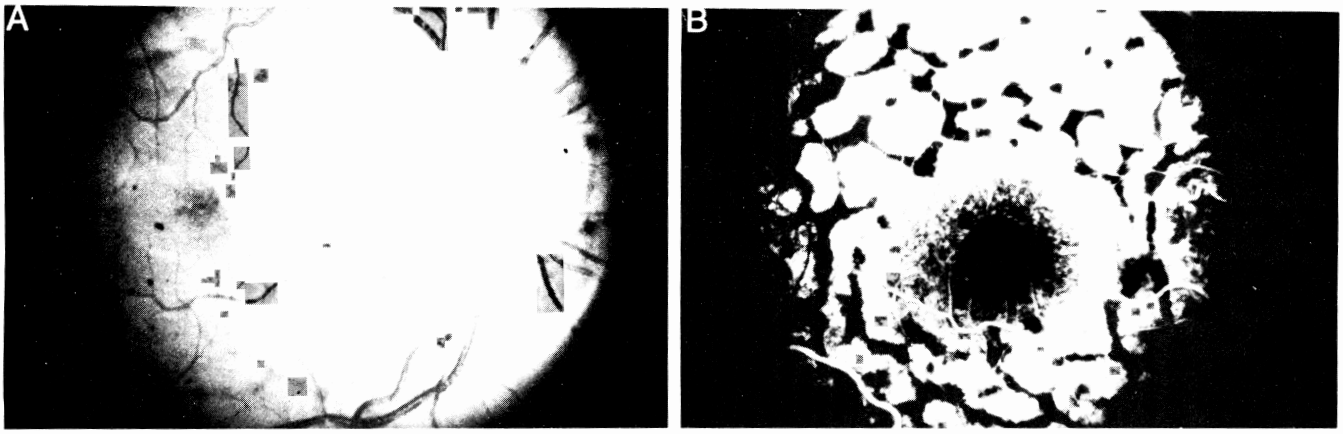


FIG 92–1.
Reticular dystrophy: fundus photograph **(A)** and fluorescein angiogram **(B).**

phy increases the risk of age-related decompensation.

Fluorescein angiography is extremely important in the evaluation of pattern dystrophies since the RPE changes are often difficult to see on ophthalmoscopic examination (see Figs 92–1 to 92–3). A fundus that shows only mild nonspecific pigment epithelial changes on direct examination may display striking granular or pigmentary patterns on angiography that extend more peripherally than expected. The pigmentary patterns visible on angiography are not associated with leakage of dye through the pigment epithelium or with secondary changes such as choroidal neovascularization (except in elderly patients[13] or two isolated cases[2] in which pattern dystrophy might not be the real cause).

PHYSIOLOGICAL FINDINGS

By definition, the pattern dystrophies are associated with relatively little functional deficit. We have already noted that visual acuity may be mildly reduced, depending upon the degree of foveal involvement, and there may be a mild degree of contrast sensitivity loss or color vision disturbance.[5] The visual fields and dark adaptation have been normal in the vast majority of reported cases, although individual pedigrees with central sensitivity loss[3] and subnormal dark adaptation[10] have been described. Severe loss of acuity should make one think of other disorders such as Stargardt's disease, vitelliform dystrophy, or age-related macular degeneration.

The electrophysiological findings in pattern dys-

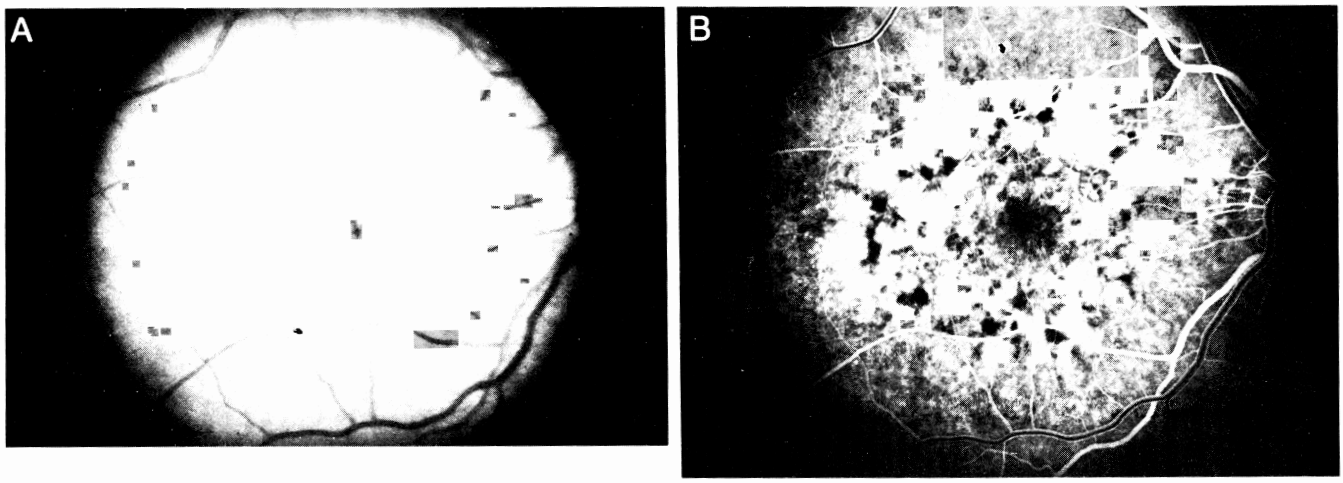
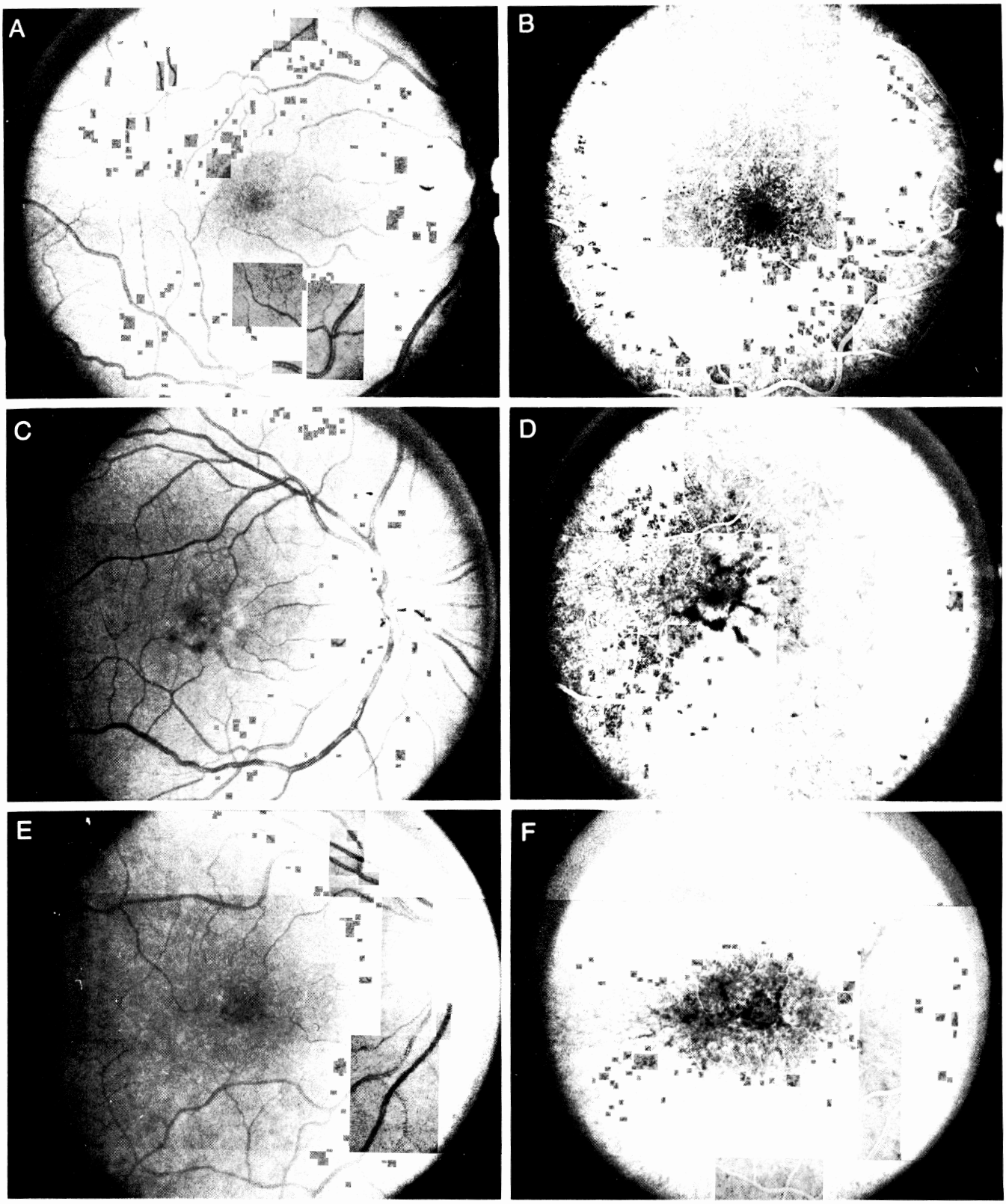


FIG 92–2.
Pattern dystrophy in a 29-year-old female with 20/25 visual acuity. Her electro-oculogram (EOG) light/dark ratios were 2.9 OD and 4.1 OS; her electroretinogram (ERG) is shown in Figure 92–4.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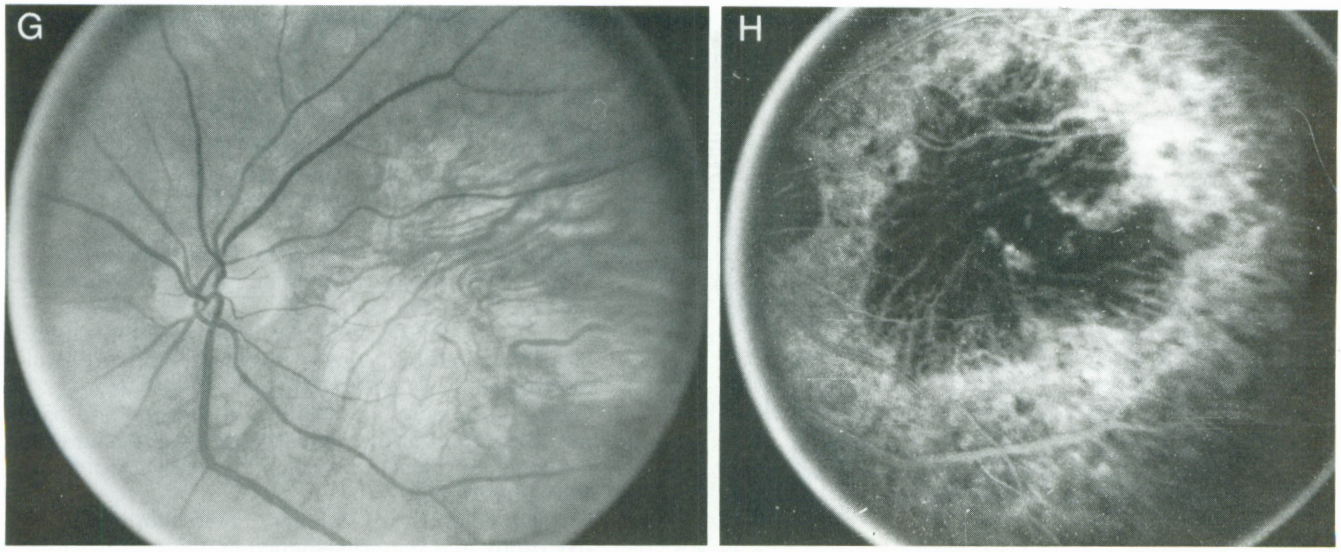

FIG 92–3.
Different patterns of pigmentation in a family with pattern dystrophy. Fundus photographs on the *left,* fluorescein angiograms on the *right.* **A** and **B,** 15-year-old daughter with granular pigmentation; visual acuity, 20/20. **C** and **D,** 20-year-old son with local reticular pigmentation; visual acuity, 20/25. **E** and **F,** 43-year-old mother with irregular reticular pigmentation; visual acuity, 20/25 to 20/30. **G** and **H,** 80-year-old sister of the grandmother; atrophic macular degeneration and a suggestion of pigmentary abnormalities in the surrounding areas are present (visual acuity, count fingers at 10 ft).

trophy are essentially limited to the EOG. The ERG has universally been reported as normal (Fig 92–4), while the EOG (which might be expected to reflect pigment epithelial abnormality) is either normal or only modestly subnormal.[1, 3, 4, 9, 10, 13, 18, 19] Individual cases with very low light/dark ratios have been noted, but most commonly the ratio is in the 1.5 to 1.8 range if suppressed at all (see Fig 92–2 and 92–5). The EOG findings are quite distinct and distinguishable from those in vitelliform dystrophy in which every affected family member (regardless of fundus lesions) has an extremely reduced light/dark

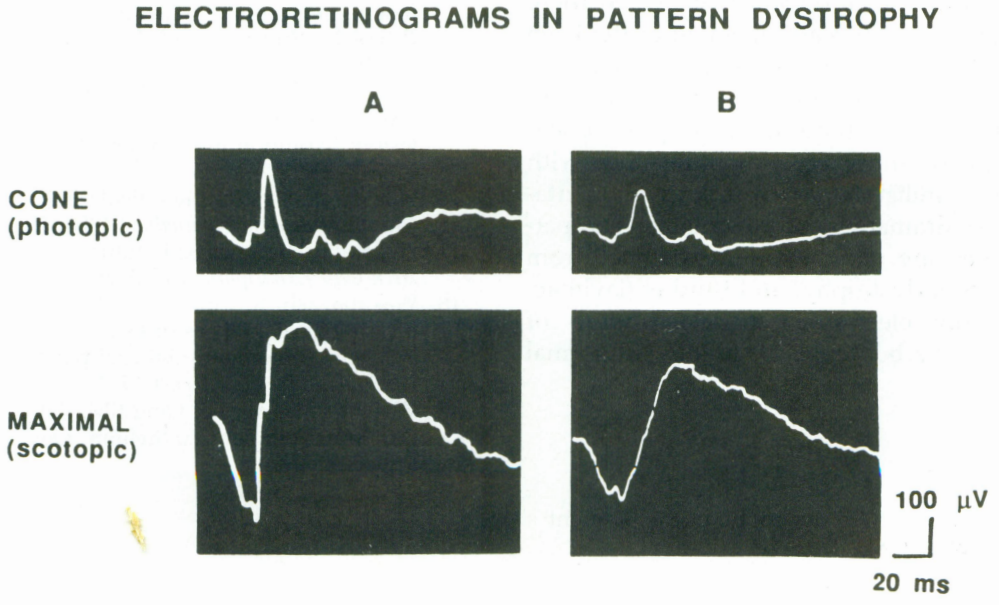

FIG 92–4.
ERGs in two cases of pattern dystrophy. **A,** the patient in Figure 92–2. **B,** the mother in Figure 92–3.

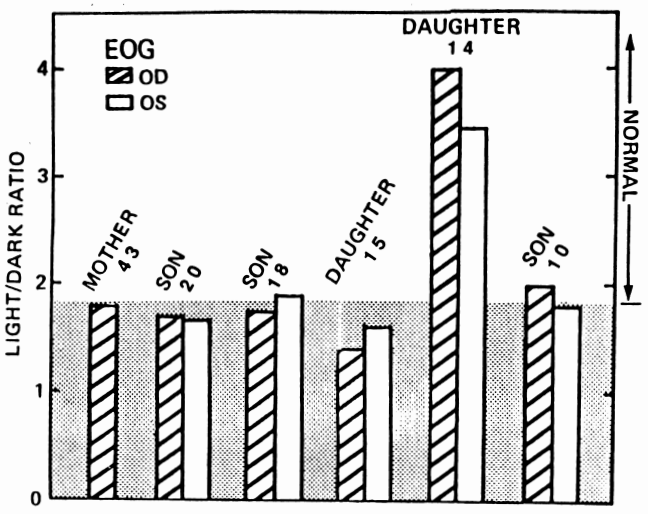

FIG 92–5.
EOG light/dark ratios in a family with pattern dystrophy (see Fig 92–3). All of the affected members had borderline values. The daughter with a high ratio had a normal-appearing fundus.

ratio. The hyperosmolarity response has been found to be abnormal in one patient with pattern dystrophy who had a normal EOG,[18] which suggests that the hyperosmolarity response may be a more sensitive indicator of diffuse RPE abnormality.

SUMMARY

The pattern dystrophies represent a group of inherited pigment epithelial disorders in which there is pigment clumping and rearrangement at the level of the RPE in the macula and/or periphery, with little or no loss of acuity or retinal sensitivity. This includes disorders such as butterfly dystrophy, reticular dystrophy, and fundus pulverulentus along with patients whose familial pattern of fundus pigmentation may be less dramatic or more variable. The pattern dystrophies are clinically quite distinct from drusen, vitelliform dystrophy, and fundus flavimaculatus. The only electrophysiological finding of some constancy is a borderline or mildly subnormal EOG.

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