Case Report: Congenital Drusen of the Retina and Hypertrophy of the Retinal Pigment Epithelium in the Same Eye

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The patient is a 24 year old caucasian female with no history of ocular or systemic disease and normal visual acuity in each eye. The right eye was unremarkable, both centrally and peripherally. In the left foveal region a number (roughly a dozen) of orange-colored, discrete circular spots were seen in the deep retina (Fig. 1, upper). These spots showed a tendency to glow in retro- or indirect illumination. Their diameter was on the order of 30–40 micrometers. These findings are consistent with retinal drusen. In the area superior to the fovea, a number of similar-sized spots of increased RPE pigmentation were seen, in proximity to the branches of the temporal retinal vessels.

In the inferior retina of the same eye, slightly posterior to the equator, a number of large pigmented areas were found (Fig. 1, lower). The shape (elliptical, with the long axis pointing toward the posterior pole), size, color and flatness of these pigmented areas, coupled with the absence of any corresponding scotomas are consistent with hypertrophy of the retinal pigment epithelium[1]. These pigmented lesions are benign. While perimetric testing of the left eye revealed no abnormalities, there were a few, rather variable disturbances in the Amsler grid field of the left eye: these consisted of one or two areas (each roughly one degree in diameter) of slight blurring of the lines within one degree of the fixation point. There were no scotomas.

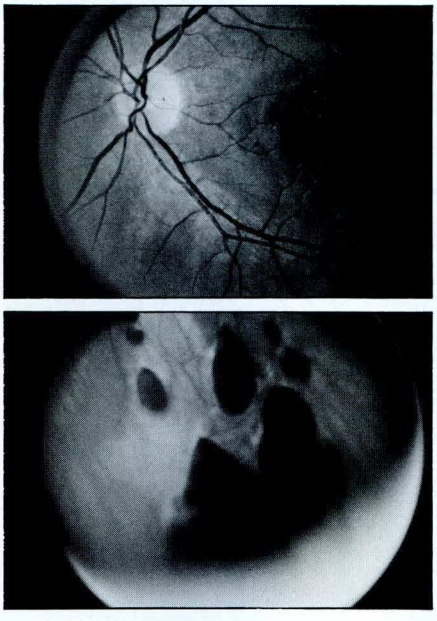

FIGURE 1

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Comment:

The role of the retinal pigment epithelium (RPE) in the genesis of localized thickenings of Bruch's membrane (drusen) has been described in detail by Farkas et al[2], Krill et al[3], Gass[4], and more recently by Ishibashi et al[5]. These authors all agree that the basis of retinal drusen (whether of the congenital or acquired variety) is some degree of abnormal RPE function, leading to deposition of hyaline-like material on the underlying Bruch's membrane.

In his longitudinal study of 200 patients with drusen and disciform macular detachment and degeneration, Gass[4] noted that approximately 33% of the patients showed large pigment epithelial cells in the equatorial area of the fundus, frequently adjacent to retinal drusen. The present case appears to show a corresponding appearance at the posterior pole.

In the last twenty years, retinal drusen have been found in association with a surprisingly wide range of ocular and systemic anomalies. These include: drusen of the optic nerve head[3], peripheral areas of enlargement of RPE cells[4], and pseudoxanthoma elasticum[3,6]. Drusen of the nerve head, in turn, have been found in association with intracranial tumors (such as craniopharyngioma and chromophobe adenoma)[7], mesodermal dysgenesis of the optic nerve head[8], and minimal brain dysfunction (clumsiness, delayed speech development, learning difficulties later in life)[9].

The findings of this case serve to remind the clinician that the retinal pigment epithelium may participate in formation not only of retinal drusen but also of sometimes dramatic (but in this case benign) peripheral pigmented areas.

References

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