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A PIGMENTED FUNDUS LESION WITH PSEUDOHYPOPYON AND SUBRETINAL FLUID RESEMBLING UNILATERAL RETINAL PIGMENT EPITHELIUM DYSGENESIS

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Purpose: To describe multimodal imaging findings in a patient with a rare, symptomatic fundus lesion arising from the retinal pigment epithelium.

Methods: Case report.

Results: A 36-year-old woman presented with photopsia in her left eye. Funduscopy revealed an 8-mm × 7-mm, dark brown lesion at the level of the retinal pigment epithelium inferior to the macula. The lesion had an irregular, cauliflower-like border and a light grey subretinal pseudohypopyon. On fundus autofluorescence, the lesion was markedly hypoautofluorescent with an irregular hyperautofluorescent margin. It was generally hyopofluorescent on fluorescein angiography and moderately hypofluorescent on indocyanine green angiography. Spectral-domain optical coherence tomography revealed a fine layer of subretinal fluid over the entire lesion, thinning of the outer retinal layers with loss of photoreceptors, and an irregular retinal pigment epithelium. Multiple drusen-like subretinal deposits were located along the lesion margin, and inferiorly, the pseudohypopyon appeared as a hyperreflective subretinal mass. During 3 years of follow-up, her symptoms remained unchanged and fundus photography showed minimal enlargement of the lesion.

Conclusion: Multimodal imaging findings of a solitary pigmented retinal pigment epithelium lesion with pseudohypopyon and subretinal fluid are shown. The lesion may represent an atypical variant of unilateral retinal pigment epithelium dysgenesis.

RETINAL CASES & BRIEF REPORTS 18:101–105, 2024

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Benign pigmented fundus lesions are commonly found during routine eye examinations. Some of these lesions arise from the retinal pigment epithelium (RPE) and include conditions such as congenital hypertrophy of the RPE (CHRPE), unilateral RPE dysgenesis (URPED), torpedo maculopathy, congenital simple hamartoma of the RPE, combined hamartoma of the retina and RPE, adenoma of the RPE, and reactive RPE hyperplasia.

Unilateral RPE dysgenesis is characterized by a unilateral, solitary, and usually peripapillary located lesion with central atrophy, peripheral fibrosis and hyperplastic changes of the RPE. The lesion is surrounded by an irregular, pathognomonic scalloped margin with focal hyperpigmentation of the RPE, leading to a typical leopard-spot appearance.[1] Solitary CHRPE typically presents as a round, flat, sharply demarcated, and darkly pigmented lesion in the midperipheral fundus, often with depigmented lacunae and a marginal halo.[2] For both conditions, structural alterations of the RPE and the overlying retinal layers

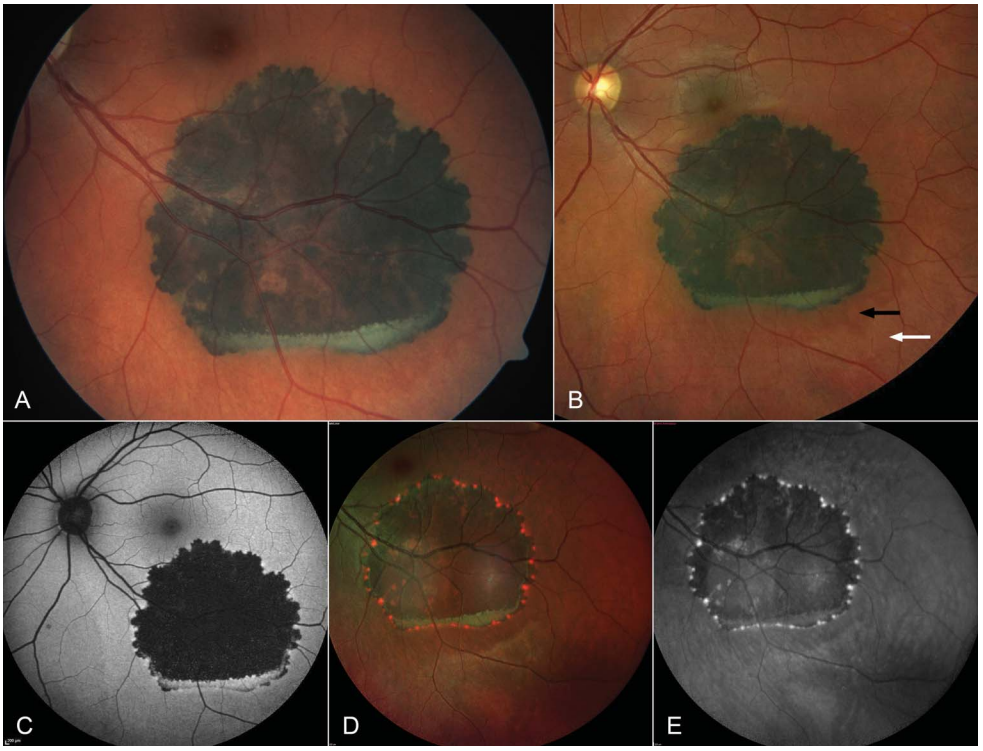

Fig. 1. A–E. Multimodal imaging of the patient's left eye. **A.** Fundus photograph at initial presentation, showing a darkly pigmented lesion with a light grey subretinal pseudohypopyon. **B.** Wide-angle fundus photograph taken about 2 years after the first visit, showing a minimal increase in lesion size, especially nasally. Note the surrounding white without pressure (white arrow) and dark without pressure (black arrow) fundus abnormalities. **C.** Fundus autofluorescence image demonstrating hypoautofluorescence of the lesion and irregular hyperautofluorescence of its margin and pseudohypopyon. **D.** Multicolor composite image showing multiple bright red spots along the lesion margin. **E.** Infrared reflectance image where the same spots appear highly reflective.

can lead to visual field defects, and over time, subtle enlargement of the lesions may occur.[2,3]

Herein, we report an unusual, pigmented fundus lesion with some similarities to URPED and CHRPE, and discuss the differential diagnostic considerations.

Case Report

A 36-year-old, previously healthy Caucasian woman had noticed photopsia in the form of flickering and shimmering lights, and slight photosensitivity in her left eye over the past six months. She was in good general health with no history of ocular disease or trauma, neither was there any family history of ocular or systemic disease. Her uncorrected visual acuity was 20/16 in both eyes, the intraocular pressures were within normal limits, and slit-lamp examination showed normal anterior segment structures. There were no signs of inflammation, neither in the anterior nor in the posterior segments. Examination of her right eye was unremarkable. Funduscopy of the left eye revealed a darkly pigmented lesion of about 8 mm × 7 mm with a fine layer of overlying subretinal fluid (SRF), located at the inferior temporal vascular arcade and extending toward the central macular area. The lesion had a dark brown color and a very irregular, almost cauliflower-like, distinct border. A light grey, granular, subretinal pseudohypopyon of 0.7 mm was observed inferiorly within the boundary of the pigmented lesion (Figure 1A). Below the lesion, there was a zone of slightly darker fundus surrounded by a lighter area, similar to white and dark without pressure abnormalities (Figure 1B).

Fundus autofluorescence demonstrated marked hypoautofluorescence of the entire lesion, with irregular hyperautofluorescence of its margin and the pseudohypopyon (Figure 1C). On the multicolor composite image, the lesion was surrounded by multiple bright red spots (Figure 1D), which also appeared highly reflective on the infrared reflectance image (Figure 1E). The blue and green reflectance images were unremarkable. Fluorescein angiography showed a normal retinal vasculature overlying the pigmented lesion, which was generally hypofluorescent with a few patches of window defects corresponding to less pigmented areas (Figure 2, A and B). Indocyanine green angiography demonstrated normal choroidal circulation outside the lesion, which appeared moderately hypofluorescent with dense hypofluorescence of the pseudohypopyon (Figure 2C). On the late phases of indocyanine green angiography, multiple hypofluorescent spots surrounded by a thin isofluorescent ring were visible along the margin of the lesion (Figure 2D). Spectral-domain optical coherence tomography (SD-OCT) through the lesion revealed thinning of the outer retinal layers with loss of photoreceptors and an irregular and slightly thickened RPE, leading to variable optical transmission. A thin layer of SRF was located over the entire lesion (Figure 3A). Inferiorly, the pseudohypopyon was observed as a hyperreflective subretinal mass (Figure 3B). Multiple drusen-like subretinal deposits at the level of the RPE were located along the entire lesion margin (Figure 3C), matching the hyperreflective spots visible on the multicolor and infrared reflectance images. Beyond the inferior border of the lesion, the spectral-domain-OCT scans

None of the authors has any financial/conflicting interests to disclose.

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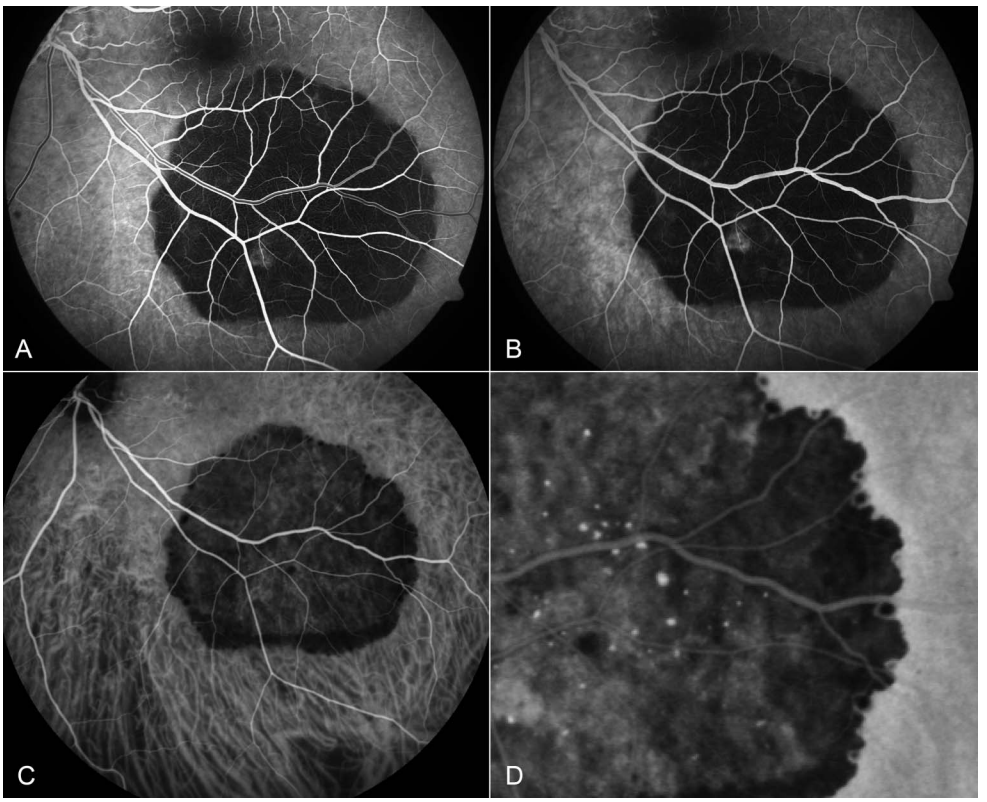

Fig. 2. A–D. Angiographic images of the patient's left eye. **A.** Early phase fluorescein angiogram demonstrating normal retinal arterial and capillary circulation over the hypofluorescent lesion. **B.** Late phase fluorescein angiogram revealing a few, slightly hyperfluorescent window defects. **C.** Early phase indocyanine green angiogram demonstrating normal choroidal circulation outside the lesion, which appears moderately hypofluorescent with dense hypofluorescence of the pseudohypopyon. **D.** Late-phase indocyanine green angiogram revealing multiple hypofluorescent spots surrounded by a thin isofluorescent ring along the lesion margin.

revealed hyporeflectivity of the ellipsoid and interdigitation zones (Figure 3C), consistent with the area of "dark without pressure" observed by funduscopy. The choroid layer underneath the lesion seemed normal. B-scan ultrasonography showed a lesion with high internal reflectivity and a thickness of about 1 mm (Figure 4A).

Based on the clinical and multimodal imaging findings, the condition was considered to be a benign RPE lesion, and the patient was scheduled for observation with semi-annual follow-up. During the following years, repeated examinations documented subtle, flat enlargement of the pigmented lesion (Figure 1B). At the last follow-up visit, 3 years after her initial presentation, her symptom of photopsia remained largely unchanged and the uncorrected visual acuity was still 20/16 in both eyes. Visual field testing of the left eye demonstrated a small, paracentral scotoma corresponding to

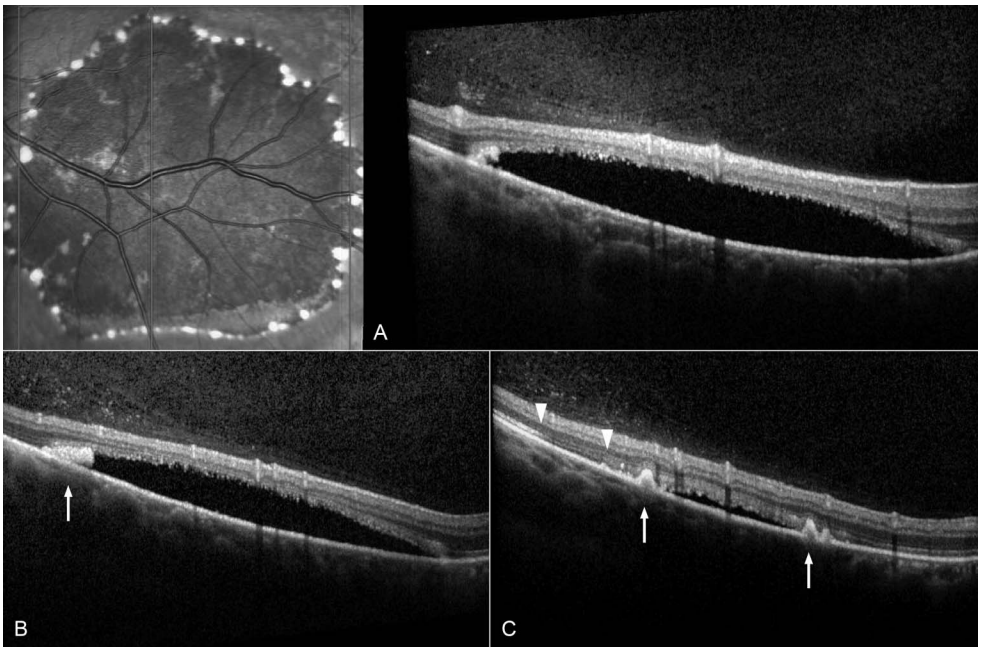

Fig. 3. A–C. Spectral-domain optical coherence tomography scans of the patient's left eye. **A.** Vertical scan through the center of the lesion showing thinning of the outer retinal layers, loss of photoreceptors and irregularity of the retinal pigment epithelium. Note the subretinal fluid and the slight elevation of the overlying retina. **B.** Vertical scan through the subretinal hyperreflective material of the pseudohypopyon (white arrow). **C.** Vertical scan through the temporal lesion margin revealing drusen-like subretinal deposits (white arrows). Note the loss of the ellipsoid and interdigitation zones (between the two white arrowheads) corresponding to the area of "dark without pressure" inferior to the lesion.

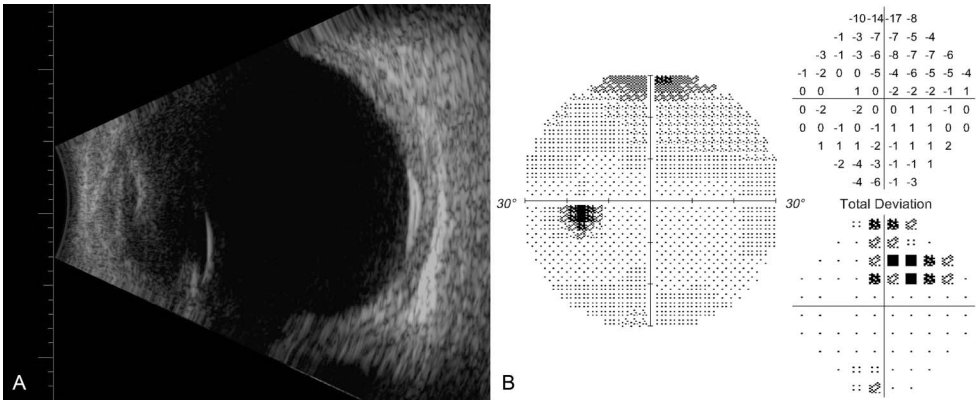

Fig. 4. A. B-scan ultrasonogram of the patient's left eye showing a minimally elevated, acoustically solid mass with a thickness of about 1 mm. **B.** Results of Humphrey central visual field test of the patient's left eye demonstrating a small scotoma above the fixation point, corresponding to the location of the pigmented fundus lesion.

the site of the lesion (Figure 4B). Fundus photographs of the left eye revealed a minimal and symmetrical increase in lesion size. The patient is still under regular follow-up.

Discussion

Most RPE lesions can be reliably diagnosed based on their clinical presentation and characteristic imaging findings. Although the funduscopic appearance of the present lesion was markedly different from other known conditions of the RPE, it had some imaging characteristics in common with URPED and CHRPE. In particular, the deep hypoautofluorescence of the lesion and its irregular hyperautofluorescent margin are consistent with the findings in URPED.[1,3] The irregular border of the present lesion may also resemble that of URPED, although closer inspection revealed some significant differences. The present lesion lacked the thin marginal fringes of mild fibrosis and atrophy that extend into the hyperpigmented RPE leading to the characteristic leopard-spot pattern observed in URPED. Another unusual finding in our patient was the drusen-like subretinal deposits located along the lesion margin. Focal thickening of the RPE is not a typical finding in URPED, but in a recently reported case, spectral-domain-OCT revealed RPE alterations in the form of hyperplasia and possible shallow RPE detachments.[4] In the present case, the outer retinal thinning and hypofluorescence on fundus autofluorescence, fluorescein angiography, and indocyanine green angiography are similar to what is observed in solitary CHRPE.[5,6] The surrounding white and dark without pressure fundus abnormalities have also recently been associated with CHRPE.[7,8] However, in contrast to our case, CHRPE usually presents with distinct, smooth margins and overlying retinal

Table 1. Comparison Between the Present Case and Similar Fundus Lesions Regarding the Clinical and Imaging Features

	Present Lesion	URPED	CHRPE
Number	Solitary	Solitary	Solitary or multiple
Location	Paracentral	Usually peripapillar	Usually midperipheral
Laterality	Unilateral	Unilateral	Unilateral or bilateral
Progressive	Yes	Yes	Yes
Symptoms	Photopsia	Visual loss if macula involved	Visual loss if macula involved
Visual field defect	Yes	Yes	Yes
Lesion shape	Round	Round or geographic	Round or geographic
Lesion border	Cauliflower-like	Scalloped or fringe-like	Sharply demarcated
Pigmentation	Hyperpigmented	Variable, leopard-spot pattern	Hyperpigmented with lacunae
Pseudohypopyon	Yes	No	No
SD-OCT	Retinal thinning, irregular and slightly thickened RPE	Thickened and disorganized retina, irregular RPE	Retinal thinning, irregular and thickened RPE
Subretinal fluid	Yes	No	Subretinal cleft
Subretinal drusen-like deposits	Yes	No	No
FAF	Hypoautofluorescent with hyperautofluorescent margin	Hypoautofluorescent with hyperautofluorescent margin	Hypoautofluorescent with isoautofluorescent lacunae
FA	Hypofluorescent	Hyperfluorescent	Hypofluorescent
ICGA	Hypofluorescent	Normal	Hypofluorescent

FA, fluorescein angiography; FAF, fundus autofluorescence; ICGA, indocyanine green angiography.

vascular abnormalities.[2,9] A comparison between the present case and the aforementioned fundus lesions is presented in Table 1.

Among the most striking features of the present case were the subretinal pseudohypopyon, the drusen-like subretinal deposits, and the amount of SRF. The light grey and hyperautofluorescent pseudohypopyon may represent subretinal accumulation of shed photoreceptor outer segments and lipofuscin-containing debris from degenerated RPE cells. The drusen-like deposits were clearly visible on multicolor imaging, indocyanine green angiography, and spectral-domain-OCT. Subretinal fluid is usually not observed in URPED, CHRPE, or other similar RPE lesions. However, in a study on 18 patients with CHRPE, Fung et al[5] found a thin rim of fluid between the retina and the pigmented portion of the lesion in six patients. The authors chose the term "subretinal cleft" rather than SRF because the fluid seemed to be located within the defect of absent or thinned outer retinal tissue. Golchet et al[10] observed a similar subretinal cleft in patients with torpedo maculopathy. The amount of SRF in the present case was larger, and unlike the cases described by Fung et al[5] and Golchet et al,[10] it led to a slight elevation of the overlying retina.

In summary, we describe the multimodal imaging findings of a symptomatic, solitary RPE lesion with pseudohypopyon and SRF, sharing certain features with URPED. Whether the lesion should be considered as an atypical variant of URPED or a distinct entity remains unclear.

Key words: pigmented fundus lesion, multimodal imaging, differential diagnosis, unilateral retinal pigment epithelium dysgenesis, congenital hypertrophy of the retinal pigment epithelium.

Acknowledgments

There are no sponsoring organizations or grants to acknowledge.

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