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CASE REPORT

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An atypical form of retinitis pigmentosa: A case report

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Key Clinical Message

This case contributes to the new knowledge of unilateral pericentral RP. Multimodal imaging provides valuable information to better understand the disease. The development is significantly slow and defines a better clinical course than typical RP. Pericentral RP presents with unremarkable night blindness, which can be easily misdiagnosed by ophthalmologists.

Abstract

Unilateral pericentral retinitis pigmentosa (RP) is a rare, atypical, mild form of RP that affects the pericentral retina close to the vascular arcades and tends to spare the far-peripheral retina in one eye, while the fellow eye remains completely unaffected. A 71-year-old Chinese woman presented with blurred vision in her right eye for several decades, however, her left eye was unaffected. Fundus photography revealed bone-spicule pigmentation located at the pericentral retina of her right eye. Spectral-domain optical coherence tomography revealed significant atrophy of the outer retina and retinal pigment epithelium, and an evident absence of the ellipsoid zone in the right eye. The visual field showed a wide-range paracentral scotoma in the right eye. The full-field electroretinogram demonstrated subnormal amplitudes of dark and light adaption in the right eye. No significant changes were observed during a further 5-year follow-up. Multimodal imaging provides valuable information to better understand unilateral pericentral RP. The development of unilateral pericentral RP is significantly slow and defines a better clinical course than the typical RP. Pericentral RP presents with slight visual reduction and unremarkable night blindness, which can be easily misdiagnosis by ophthalmologists. Reasonable prevention measures, screening methods, and timely diagnosis are beneficial for RP patients.

KEYWORDS

best-corrected visual acuity, case report, full-field electroretinogram, multimodal imaging, unilateral pericentral retinitis pigmentosa

1 | INTRODUCTION

Retinitis pigmentosa (RP) is a hereditary retinal disease caused by the degeneration of the retinal pigment epithelium and secondary photoreceptor atrophy, which is characterized by bone-spicule pigment migration located at the mid-peripheral and far-peripheral retina, night blindness, and progressive constricted visual field in both eyes.[1] It commonly occurs between the ages of 10 and 30. Several forms of RP have been described in the literature, such as typical RP, sector RP, pericentral RP, and nonpigmented RP.[2,3]

In comparison to typical RP, pericentral RP is a significantly rare, atypical, mild form of RP that affects the mid-peripheral retina and tends to spare the far-peripheral retina and usually occurs bilaterally. It does not manifest the typical clinical manifestations and fundus signs of RP.[4–6] The diagnosis of the disease is mainly based on the fundus appearance, full-field electroretinogram, spectral-domain optical coherence tomography (SD-OCT), and visual field.[4,6] However, to the best of our knowledge, unilateral pericentrtal RP has not been described in previous reports. Herein, we observed and evaluated unilateral pericentrtal RP in a Chinese woman simultaneously using multimodal imaging.

2 | CASE PRESENTATION

A 70-year-old Chinese woman presented with blurred vision in her right eye for several decades, however, her left eye was unaffected. She presented with unremarkable night blindness in her right eye. She reported no history of medication or trauma, and no other similar cases observed in her family. She also denied any ophthalmic or systemic diseases related to infection, medication, or tumor. Her best-corrected visual acuity was 20/400 OD and 20/30 OS. Fundus photography revealed scattered bone-spicule pigmentation located at the atrophic pericentral retina of her right eye, with mild attenuation of retinal vessels (Figure 1A), which was not observed in her left eye (Figure 1B). Spectral-domain optical coherence tomography (SD-OCT) revealed significant atrophy of the outer retina and retinal pigment epithelium, and an evident absence of the ellipsoid zone in the right eye (Figure 1C), thickened retinal neurosensory layer and fovea, and well-preserved ellipsoid zone were observed in the left eye (Figure 1D). The result of OCT angiography of the right eye was normal in the whole, superficial, deep, and outer retinal layers (Figure 2A–D), whereas the atrophy of choroidal capillaries and choroidal vessels was visible at the choroidal capillary layer (Figure 2E). The visual field showed a wide-range paracentral scotoma in the right eye (Figure 3A), which was normal in the left eye (Figure 3B). The full-field electroretinogram (ff-ERG) revealed subnormal amplitudes of dark and light adaption in the right eye (Dark-adapted 0.01 responses, b-wave amplitude: 91.7, normal range: 216–341 μV (Figure 4A); Dark-adapted 3.0 responses, a-wave amplitude: 185.0, normal range: 232–375 μV, b-wave amplitude: 342.7, normal range: 479–568 μV (Figure 4B); Dark-adapted 3.0 oscillatory potentials, P2 amplitude: 42.9, normal range: 77–150 μV (Figure 4C); Light-adapted 3.0 responses, a-wave amplitude: 36.3, normal range: 12–95 μV, b-wave amplitude: 75.4, normal range: 147–222 μV (Figure 4D); Light-adapted 30 Hz flicker responses, P2 amplitude:

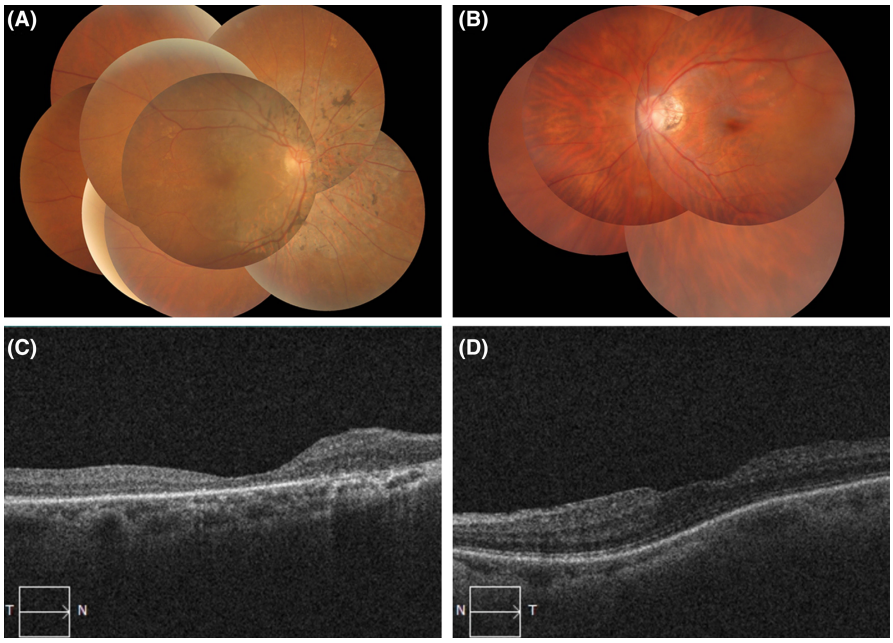

FIGURE 1 Fundus photography revealed scattered bone-spicule pigmentation located at the atrophic pericentral retina of her right eye, with a normal-appearing optic disc, and mild attenuation of retinal vessels (A), which was not observed in her left eye (B). SD-OCT revealed significant atrophy of the outer retina and retinal pigment epithelium, and an evident absence of the ellipsoid zone in the right eye (C), thickened retinal neurosensory layer and fovea, and well-preserved ellipsoid zone were observed in the left eye (D).

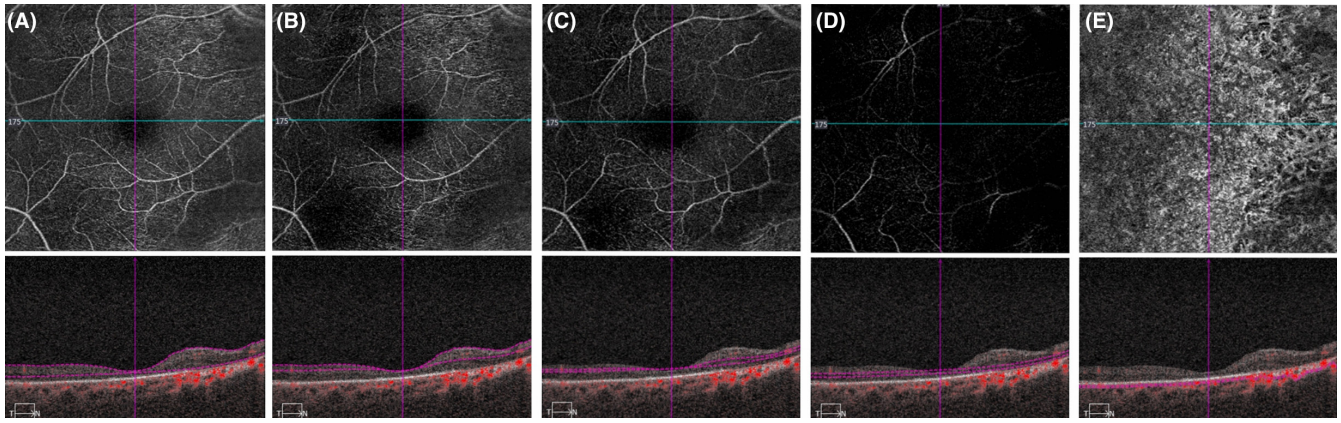

FIGURE 2 The result of OCT angiography of the right eye was normal in the whole, superficial, deep, and outer retinal layers (A–D), whereas the atrophy of choroidal capillaries and choroidal vessels was visible at the level of the choroidal capillary layer (E).

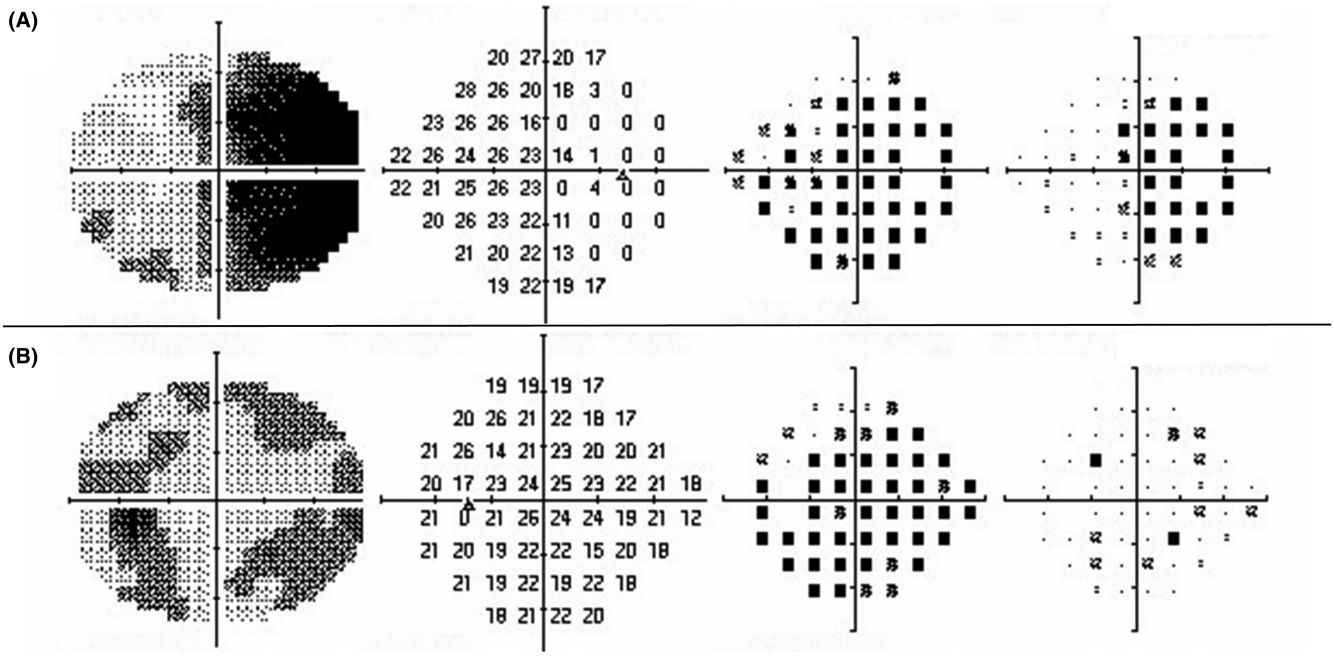

FIGURE 3 The visual field showed a wide-range paracentral scotoma in the right eye (A), which was normal in the left eye (B).

53.6, normal range: 99–171 μV (Figure 4E), which were completely normal in the left eye. Color vision revealed a moderate defect in the right eye, while was normal in the left eye. The audio-vestibular examination results were normal.

Based on the above findings, the patient was diagnosed with unilateral pericentral RP. She was closely observed, and there were no significant changes in all multimodal imaging during a 5-year follow-up.

3 | DISCUSSION

Compared to typical RP, pericentral RP is a significantly rare, atypical, mild form of RP that the patchy bone-spicule pigmentation lesions only appears to be limited in the mid-peripheral retina close to the vascular arcades.[4] It presents with unremarkable night blindness, which is distinct from typical RP.[6,8] Pericentral RP can be easily misdiagnosed by ophthalmologists. To the best of our knowledge, unilateral pericentral RP has not been described in previous reports. Some studies have confirmed that RP is related to mutations in genes, therefore, RP patients may present isolated (non-syndromic) RP as well as syndromic forms (e.g., Usher syndrome, Heimler syndrome).[1,7] However, different from typical RP, the etiology of unilateral pericentrtal RP remains to be established.

Using SD-OCT, well-preserved ellipsoid zone is observed on the central macula in typical RP, which is quite distinct from pericentral RP. Pericentral RP shows

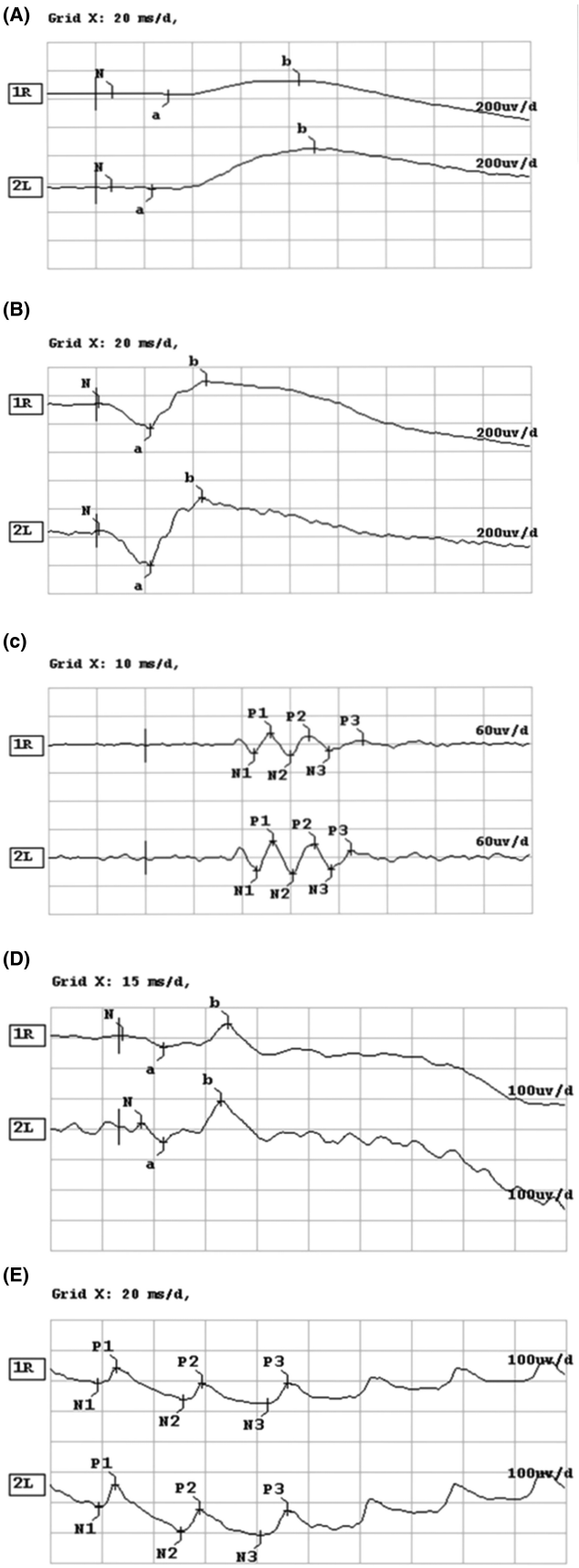

FIGURE 4 The full-field electroretinogram (ff-ERG) revealed subnormal amplitudes of dark and light adaption in the right eye (Dark-adapted 0.01 responses, b-wave amplitude: 91.7, normal range: 216–341 μV (A); Dark-adapted 3.0 responses, a-wave amplitude: 185.0, normal range: 232–375 μV, b-wave amplitude: 342.7, normal range: 479–568 μV (B); Dark-adapted 3.0 oscillatory potentials, P2 amplitude: 42.9, normal range: 77–150 μV (C); Light-adapted 3.0 responses, a-wave amplitude: 36.3, normal range: 12–95 μV, b-wave amplitude: 75.4, normal range: 147–222 μV (D); Light-adapted 30 Hz flicker responses, P2 amplitude: 53.6, normal range: 99–171 μV (E)), which were completely normal in the left eye (R: right eye; L: left eye).

interrupted or absent ellipsoid zone on the fovea instead.[9] Karali et al. identified that 15% of patients with pericentral RP had central macular thickness reduction, and all patients had interrupted or absent ellipsoid zone.[7]

OCT angiography is a noninvasive technique that can be used to evaluate microvasculature morphology and density for RP patients. The choroidal capillary layer reveals atrophy of the choroidal capillaries and vessels on OCT angiography, corresponding to the area of the lesions. Similarly, Corazza et al. determined a significant reduction in choroidal capillary density in patients with typical RP using OCT angiography.[10]

The visual field can precisely reflect retinal sensitivity. Dysfunction of the overlying retina and secondary photoreceptor degeneration are due to retinal pigment epithelium atrophy, which manifests a reduction in retinal sensitivity.[11] Different from the progressive constricted visual field of patients with typical RP, pericentral RP shows paracentral scotoma corresponding to the lesions on the fundus of the affected eyes.

Electroretinography is a sensitive method to diagnose RP and provides a valuable tool for future visual function.[12] In typical RP, ff-ERG shows the severe reduction in cone and rod responses. In pericentral RP, although the dark-adapted and light-adapted responses are both reduced, while these are still detectable easily. The results are consistent with those of Sandberg et al. and Karali et al.[4,7] In addition, a detailed study by Szamier et al. described the histopathological basis for the clinical findings in one postmortem donor eye of a patient with pericentral RP. The photoreceptors were absent in the precentral retina, however, a large number of normal cone and rod photoreceptors were found in the periphery and far periphery, which could account for the detectable electroretinogram.[6]

Furthermore, Sandberg et al. investigated the course of 18 patients with pericentral RP (aged 32–65 years) and found that pericentral RP progressed more slowly than typical RP.[4] Szamier et al. also observed that the progress of patients with pericentral RP was slight during 30 years.[6]

The fundus appearance, ff-ERG, SD-OCT, and visual field are effective tools for investigating RP.[4,6] Reasonable prevention measures, screening methods, and timely diagnosis are beneficial for typical and atypical patients. However, the treatment of RP patients remains challenging. Vasodilator agents, neuroprotectors, optogenic therapy, stem cell transplants, and gene therapy have been used for RP patients, however, no treatments mentioned are proven persistently effective.[13,14] Preventing marriage between close relatives can reduce the incidence rate of RP. Patients with night blindness should undergo timely ophthalmic examination and prompt treatment. Clinicians who are unable to access to the all mentioned imaging modalities can transfer the suspected RP patients to other eligible hospitals for making a definite diagnosis.

Differential diagnosis should be carefully established. First, audio-vestibular examination is recommended for pericentral RP to unravel the diagnosis of Usher syndrome, even in patients without hearing symptoms.[7] Second, cone-rod dystrophy is a bilateral, hereditary retinal disease that presents with reduced vision, photophobia, and nystagmus. The fundus of patients with cone-rod dystrophy is relatively normal, ff-ERG reveals a severe reduction in photopic response with a relatively normal scotopic response.[12] Third, patients with hydroxychloroquine retinopathy have a clear history of taking hydroxychloroquine, with the fundus lacking the bone-spicule pigment, and ff-ERG reveals severe cone damage with well-preserved rod response.[5] Besides, other potential causes such as certain medications, specific infections, traumatic factors, and cancer, in every patient with pigmentary retinopathy should also be ruled out.[12] The patient in this case reported no history of medication or trauma, and no other similar cases were observed in her family. She also denied any ophthalmic or systemic diseases related to infection, medication, or tumor. Moreover, her left eye was completely unaffected after a sufficiently long period of observation (more than 7 years). As a result, she was definitively diagnosed with unilateral pericentrtal RP.

However, this study had several limitations, fluorescence angiography, fundus autofluorescence, and genetic screening were not performed as they may help in better understanding of this disease.

In conclusion, this unique case report contributes to the new knowledge of unilateral pericentral RP simultaneously using multimodal imaging. Multimodal imaging provides valuable information to better understand the disease. The development is significantly slow and defines a better clinical course than the typical RP. Pericentral RP presents with unremarkable night blindness, which can be easily misdiagnosed by ophthalmologists. Reasonable prevention measures, screening methods, and timely diagnosis are beneficial for typical and atypical RP patients.

AUTHOR CONTRIBUTIONS

Yuanfeng Xue: Writing – original draft. **Bangtao Yao:** Writing – review and editing.

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CONFLICT OF INTEREST STATEMENT

The authors declare that they have no conflicts of interest.

DATA AVAILABILITY STATEMENT

The datasets presented in this study are included in the article, further inquiries can be directed to the corresponding author/s.

ETHICS STATEMENT

The study was approved by the Institutional Review Board of Nanjing Lishui People's Hospital.

CONSENT

Informed written consent had been obtained from the patient.

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