American Journal of Ophthalmology Case Reports 28 (2022) 101708

Contents lists available at ScienceDirect

American Journal of Ophthalmology Case Reports

journal homepage: www.ajocasereports.com/

Insidious progression of atrophic lesions in a case of posterior polar annular choroidal dystrophy

Risako Sone, Kousuke Noda *, Kiriko Hirooka, Michiyuki Saito, Susumu Ishida

Department of Ophthalmology, Faculty of Medicine and Graduate School of Medicine, Hokkaido University, N-15, W-7, Kita-ku, Sapporo, 060-8638, Japan

ARTICLE INFO

Keywords:
Posterior polar annular choroidal dystrophy
Case report

ABSTRACT

Purpose: To describe the clinical course of a case of posterior polar annular choroidal dystrophy (PPACD) followed for 5 years.

Observations: A 64-year-old female patient presented with blurred vision. The patient had no subjective symptoms of night blindness or visual field defects. At the initial visit, the patient's visual acuity was 20/20 in both eyes. Bilateral fundus examination revealed atrophic lesions surrounding the optic nerve head, extending to the temporal arcades in an annular pattern. Fundus autofluorescence (FAF) revealed hypoautofluorescent areas corresponding to atrophic lesions, and Goldmann perimetry revealed ring scotomas consistent with lesions in the fundus. Swept-source optical coherence tomography revealed retinal pigment epithelium atrophy, loss of the choriocapillaris, and dilation of the choroidal medium and large vessels in the atrophic area. Full-field electroretinography revealed a mild reduction in the combined rod–cone response. Laser speckle flowgraphy revealed a cold color in the posterior pole of both eyes. Based on clinical and imaging findings, the patient was diagnosed with PPACD and followed up for 5 years. At the 5-year visit, visual acuity remained unchanged, while FAF and Goldmann perimetry revealed a slight enlargement of the atrophic lesions and scotoma in both eyes, respectively.

Conclusions and Importance: In the present case, atrophic lesions insidiously progressed and resulted in a slight enlargement of the hypoautofluorescent area and scotoma over a 5-year follow-up period, indicating that PPACD is a gradually progressive dystrophy.

1. Introduction

Posterior polar annular choroidal dystrophy (PPACD) is a rare disease characterized by bilateral atrophy of the retinal pigment epithelium (RPE) and choriocapillaris surrounding the arcade vessels and optic disc, and it typically spares the fovea, optic disc, and peripheral retina.[1] Since its initial documentation by Yannuzzi in 2010,[2] a few cases have been reported and therefore the reports regarding long-term prognosis of the disease is limited. Herein, we report the clinical findings of a case of PPACD followed up for 5 years.

2. Case report

A 64-year-old female patient presented with a complaint of blurred vision over the preceding 4 months. The patient had no subjective symptoms of night blindness or visual field defects. Her medical history included overlap syndrome, interstitial pneumonia, Hashimoto's thyroiditis, hypertension, and diabetes. There was no family history of hereditary ocular diseases. At the initial visit, the patient's best-corrected visual acuity was 20/20 OU. The intraocular pressure was within the normal range, and the anterior segment slit-lamp examination was unremarkable, except for grade 1 nuclear cataracts OU. Dilated fundus examination revealed asymmetric RPE atrophy surrounding the optic nerve head and extending to the temporal arcades in an annular pattern, sparing the central macula. The atrophic changes were more extensive in the right eye than in the left eye. Although discrete pigment clumps were sporadically detected in the grayish atrophic areas, no bone spicule-like pigment changes in the peripheral retina or arteriolar attenuation was observed (Figs. 1 and 2). Fundus autofluorescence (FAF) imaging revealed hypoautofluorescence in the atrophic areas and hyperautofluorescence at the junction between the normal and atrophic retinas (Fig. 3). Goldmann perimetry (GP) revealed ring scotomas consistent with atrophic changes in the fundus, whereas concentric constriction of the visual field (I-4 isopter) was not detected (Fig. 4).

* Corresponding author. N-15, W-7, Kita-ku, Sapporo, 060-8638, Japan.
E-mail address: nodako@med.hokudai.ac.jp (K. Noda).

https://doi.org/10.1016/j.ajoc.2022.101708
Received 26 July 2022; Received in revised form 5 September 2022; Accepted 9 September 2022
Available online 21 September 2022

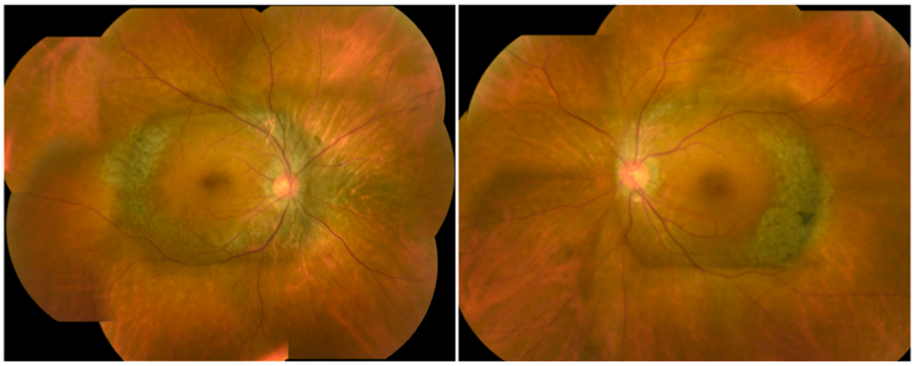

Fig. 1. Dilated fundus findings in the present case of PPACD
Dilated fundus examination revealed asymmetric RPE atrophy surrounding the optic nerve head and extending to the temporal arcades in an annular pattern, sparing the central macula. The atrophic changes were more extensive in the right eye than in the left eye. Although discrete pigment clumps were sporadically detected in the grayish atrophic areas, no bone spicule-like pigment changes in the peripheral retina or arteriolar attenuation was observed.

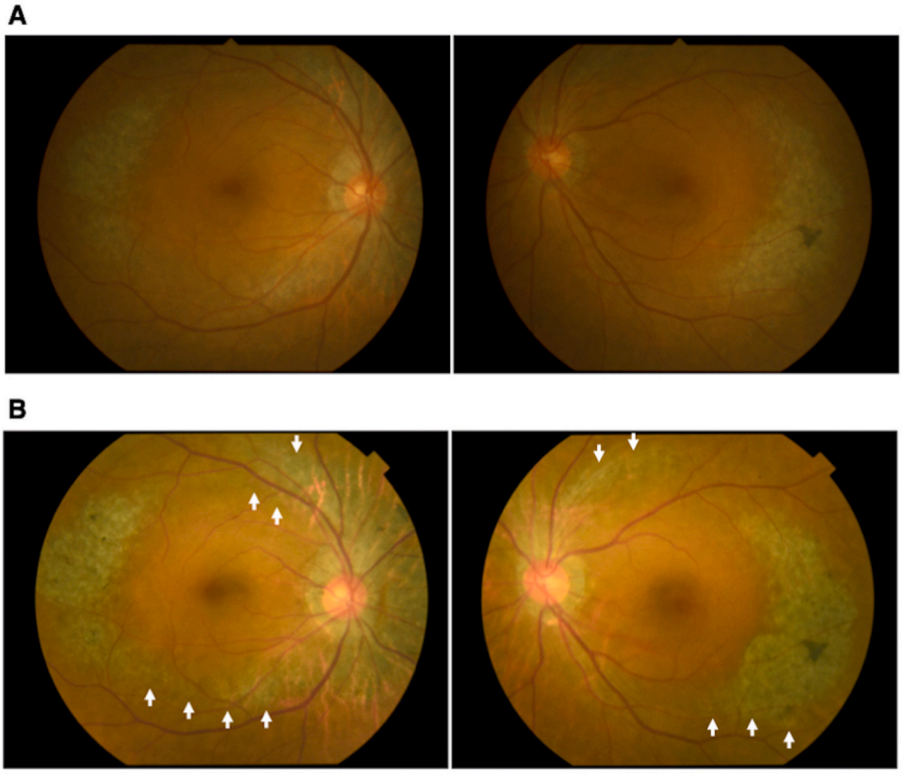

Fig. 2. Changes in fundus findings over 5 years in the present case of PPACD
Fundus images of the present case at the initial (A) and 5-year (B) visits. Slight enlargement of atrophy (arrows) was observed in both eyes.

Swept-source optical coherence tomography (SS-OCT) revealed thinning of the retina, disruption of the ellipsoid zone and RPE line, loss of the choriocapillaris, and dilation of the choroidal medium and large vessels in the atrophic area (Fig. 5). On the full-field electroretinogram (ERG), the a-wave amplitudes exhibited a mild reduction in scotopic and combined rod–cone responses in both eyes (Fig. 6). The cone and 30-Hz flicker ERGs were slightly reduced in both eyes, while both of which were within the normal range (Fig. 6). Laser speckle flowgraphy (LSFG) revealed a cold color in the posterior pole of both eyes (Fig. 7). Based on clinical and imaging findings, the patient was diagnosed with PPACD and followed up for 5 years. During the follow-up, the progression of fundus legions were not remarkable. At the 5-year visit, visual acuity remained unchanged at 20/20 in both eyes, while atrophic lesions were slightly elongated (Figs. 2 and 3) and corresponding visual field defects noted (Fig. 4) in both eyes.

3. Discussion

PPACD is a rare clinical entity characterized by ring-like atrophy surrounding the arcade vessels and optic disc. Atrophic lesions in PPACD are similar to those in retinitis pigmentosa, despite being confined to the posterior pole sparing the macula. Therefore, inherited retinal dystrophies, such as sector retinitis pigmentosa, are differential diagnoses of PPACD. However, the present case exhibited no subjective symptoms of night blindness or visual field defect. In addition, retinal degeneration was limited to the periphery of the arcade vessels and optic disc with an intact peripheral retina, and neither narrowing of the retinal vessels nor bone spicule-like pigment changes were observed in the present case. The ERG revealed a relatively favorable response, and Goldmann perimetry demonstrated ring scotomas corresponding to atrophic lesions without concentric constriction of the visual field (I-4 isopter). Apparently, the clinical characteristics of the present case were different from those of retinitis pigmentosa and allied diseases, leading to a clinical diagnosis of PPACD.

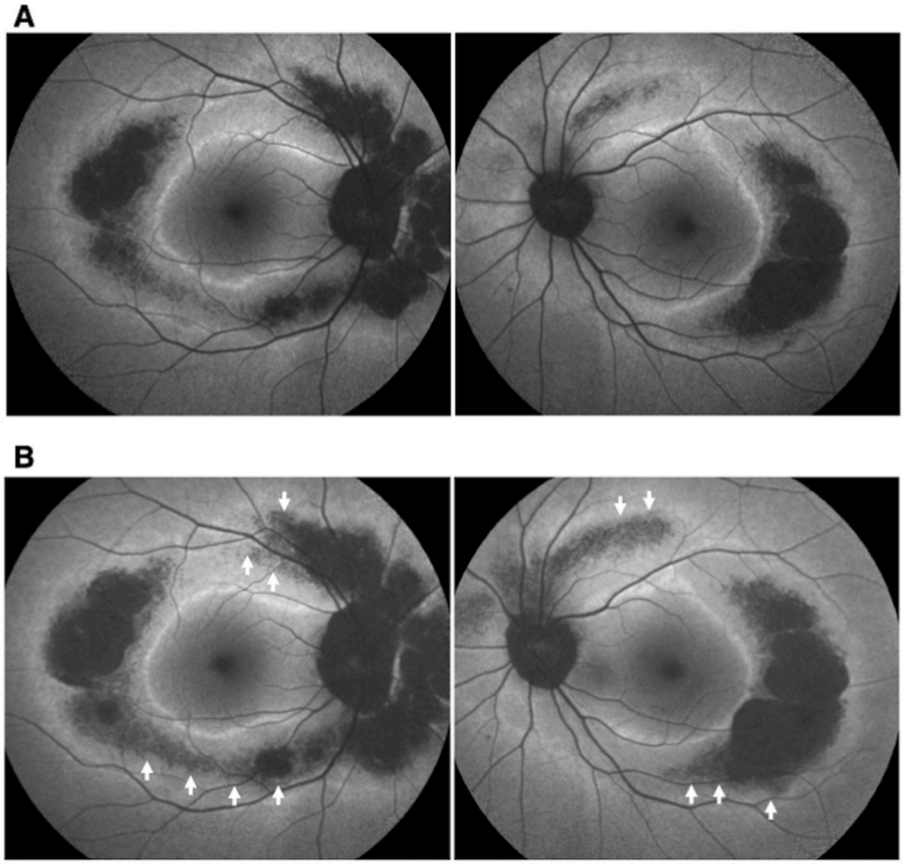

Fig. 3. Changes in FAF over 5 years in the present case of PPACD
FAF images of the present case at the initial (A) and 5-year (B) visits. FAF imaging exhibited hypofluorescence in the atrophic areas and hyperfluorescence at the junction between the normal and atrophic retina at the initial visit. Five years later, a slight enlargement of the hypofluorescence (arrows) was observed on the FAF images of both eyes.

To date, only a few cases of PPACD have been reported in the literature.[1,3–5] Of these, one case reported by Forte et al. was followed up for long-term prognosis.[3] The aforementioned PPACD case involved a 32-year-old man who exhibited an annular pattern of retinal atrophy in both eyes, similar to that in our case. After a 3-year follow-up, the previous case was reported to develop a slight enlargement of hypoautofluorescence on FAF imaging without remarkable changes in fundus findings and visual field tests. Likewise, the present case also exhibited progressive hypoautofluorescence on FAF imaging, and further progression of atrophy on funduscopic and visual field examinations was observed after 5 years of follow-up. As in the previous case, the fovea in our case was not affected during follow-up, and visual acuity remained unchanged from the initial visit. The previous and present cases indicate that PPACD is a gradually progressive disease, and FAF is useful in readily assessing the insidious progression of fundus lesions in patients with PPACD.

To the best of our knowledge, this is the first report to describe the LSFG findings in a case of PPACD. LSFG is a noninvasive method for the quantitative estimation of blood flow in the optic nerve head, choroid, and retina based on speckle statistics. The hemodynamic pattern observed on LSFG has been demonstrated to predominantly originate from the choroid (approximately 92% of the sum of the choroidal and retinal circulation),[6] with warm and cool colors in regions of increasing and decreasing blood flow, respectively. In the present case, LSFG exhibited a cold color in the posterior pole of both eyes, indicating a reduction in choroidal blood flow velocity at the macula. Notably, electrophysiological examinations clearly demonstrated a slight decrease in cone response. Previously, adaptive optics examination revealed a reduction in foveal cone photoreceptors in a case of PPACD.[3] Taken together, accumulating evidence suggests that foveal cone photoreceptors in eyes with PPACD are subclinically affected despite the intact appearance of the macula.

Intriguingly, SS-OCT visualized dilated choroidal large vessels predominantly in the atrophic retina in the present case. Although not mentioned, previous reports have also revealed dilated choroidal large vessels in OCT images.[4,5] While the pathophysiology accounting for the dilation of choroidal large vessels remains unclear, the decreased choroidal blood flow velocity detected in LSFG indicated that the dilated choroidal vessels were consequence of the congestion of choroidal blood flow. It is possible that the choroidal blood vessels are dilated due to the atrophy of the surrounding tissues or vascular wall components per se in the atrophic lesions. Alternatively, it is also possible that low-grade inflammatory changes caused the dilation of choroidal blood vessels in the atrophic lesions. Nonetheless, our case report is limited by the challenges common to studying rare diseases and the LSFG findings in cases of PPACD has not been reported so far. Therefore, further case accumulation is warranted to elucidate the pathogenesis of PPACD, including the dilation of choroidal large vessels, pathological role of choroidal blood flow on latent impairment of cone photoreceptors, and insidious progression of atrophic lesions.

4. Conclusions

In the present case of PPACD, 5-year follow-up evidently demonstrated the progression of fundus lesions, resulting in deterioration of the visual field without subjective visual disturbance, indicating that PPACD is a gradually progressive disease. Further accumulation of cases is required to elucidate the long-term prognosis and pathological mechanisms of this disease.

Patient consent

Informed consent was obtained from the patient for the purpose of publication.

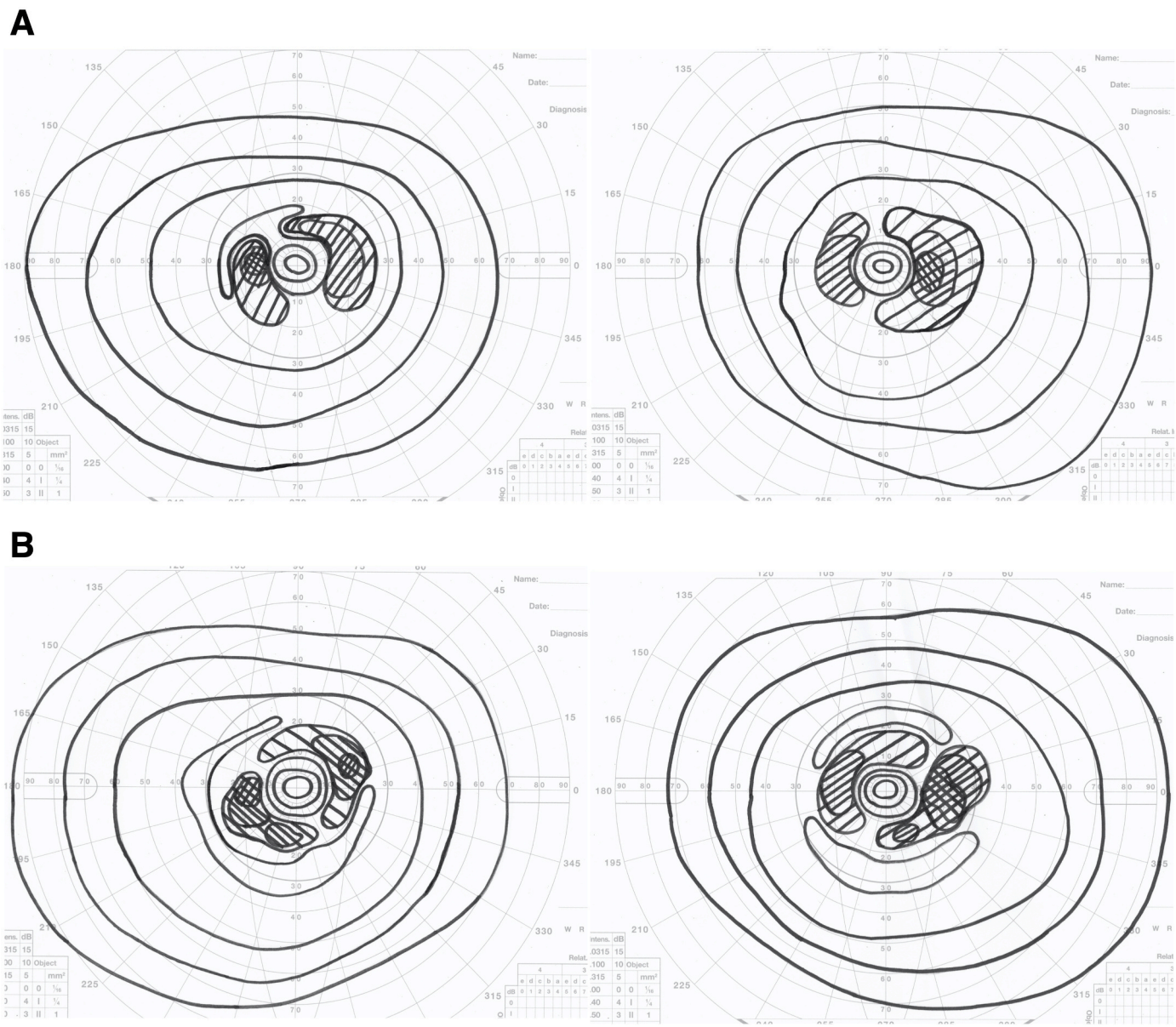

Fig. 4. Changes in GP over 5 years in the present case of PPACD
GP revealed ring scotomas consistent with atrophic changes in the fundus, whereas concentric constriction of the visual field (I-4 isopter) was not detected at the initial visit (A). Five years later, a slight enlargement of the scotoma was observed on GP in both eyes (B).

Funding

No funding or grant support.

Intellectual property

We confirm that we have given due consideration to the protection of intellectual property associated with this work and that there are no impediments to publication, including the timing of publication, with respect to intellectual property. In so doing we confirm that we have followed the regulations of our institutions concerning intellectual property.

Research ethics

We further confirm that any aspect of the work covered in this manuscript that has involved human patients has been conducted with the ethical approval of all relevant bodies and that such approvals are acknowledged within the manuscript.

IRB approval was obtained (required for studies and series of 3 or more cases).

Written consent to publish potentially identifying information, such as details or the case and photographs, was obtained from the patient(s) or their legal guardian(s).

Authorship

All listed authors meet the ICMJE criteria.
We attest that all authors contributed significantly to the creation of this manuscript, each having fulfilled criteria as established by the ICMJE.

We confirm that the manuscript has been read and approved by all named authors.

We confirm that the order of authors listed in the manuscript has been approved by all named authors.

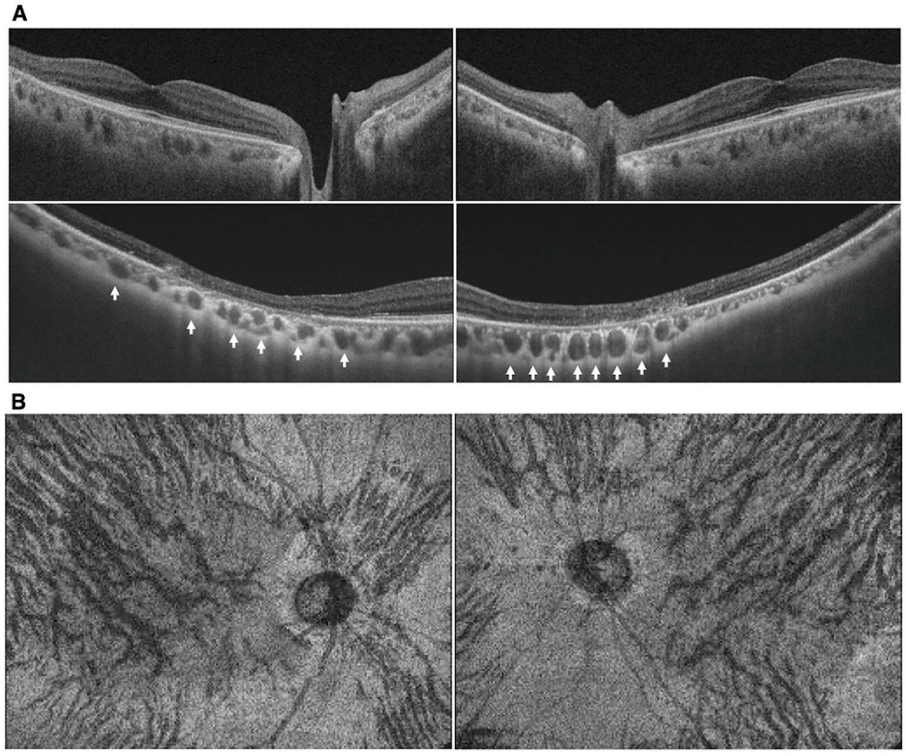

Fig. 5. SS-OCT imaging in the present case of PPACD
(A) SS-OCT revealed thinning of the retina, disruption of the ellipsoid zone and RPE line, loss of the choriocapillaris, and dilation of the choroidal medium and large vessels in the atrophic area of both eyes (arrows). (B) The SS-OCT map enfaced at the RPE level revealed dilation of choroidal medium and large vessels.

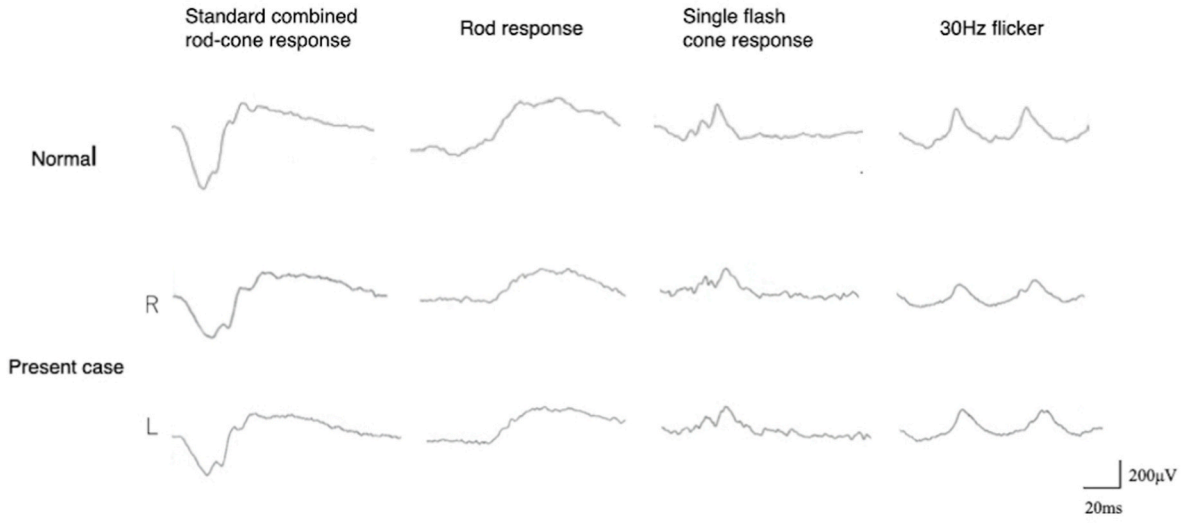

Fig. 6. Full-field ERG of the present case of PPACD
On a full-field electroretinogram (ERG), the a-wave amplitudes revealed a mild reduction in scotopic and combined rod–cone responses in both eyes. The cone and 30-Hz flicker ERGs were slightly reduced in both eyes.

Contact with the editorial office

This author submitted this manuscript using his/her account in EVISE.

We understand that this Corresponding Author is the sole contact for the Editorial process (including EVISE and direct communications with the office). He/she is responsible for communicating with the other authors about progress, submissions of revisions and final approval of proofs.

We confirm that the email address shown below is accessible by the Corresponding Author, is the address to which Corresponding Author's EVISE account is linked, and has been configured to accept email from the editorial office of American Journal of Ophthalmology Case Reports:

CRediT authorship contribution statement

Risako Sone: Investigation, Writing – original draft. **Kousuke Noda:** Conceptualization, Writing – original draft. **Kiriko Hirooka:**

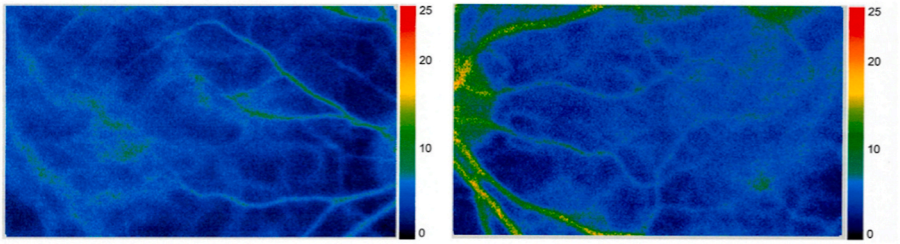

Fig. 7. LSFG in the present case of PPACD
LSFG revealed a cold-color pattern in the posterior pole of both eyes.

Investigation, Visualization, Data curation. **Michiyuki Saito:** Writing – review & editing. **Susumu Ishida:** Writing – review & editing.

Declaration of competing interest

No conflict of interest exists.

Acknowledgements

We thank the patient for granting permission to publish this information.

References

1. Lenis TL, Klufas MA, Randhawa S, Sharma M, Sarraf D. Posterior polar annular choroidal dystrophy: a case series. *Retin Cases Brief Rep.* 2017;11(Suppl 1):S24–S27.
2. Yannuzzi LA. *The Retinal Atlas.* Elsevier Health Sciences; 2010.
3. Forte R, Aptel F, Feldmann A, Chiquet C. Multimodal imaging of posterior polar annular choroidal dystrophy. *Retin Cases Brief Rep.* 2018;12:29–32.
4. Gala JM, Narayanan R. Posterior polar annular choroidal dystrophy. *BMJ Case Rep.* 2019;12, e230495.
5. Narayanan R. Posterior polar annular and hemispheric choroidal and retinal dystrophy: optical coherence tomographic angiography description of a rare case. *Indian J Ophthalmol.* 2018;66:1874–1876.
6. Sugiyama T. Basic technology and clinical applications of the updated model of laser speckle flowgraphy to ocular diseases. *Photonics: MDPI.* 2014;1:220–234.