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Posterior polar annular choroidal dystrophy: a synopsis of choroidal dystrophies impacting the central macula through a case report

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ABSTRACT

Posterior polar annular choroidal dystrophy (PPACD) is an uncommon disease characterized by atrophy of the retinal pigment epithelium and choriocapillaris in an annular pattern. To date, only a limited number of cases has been reported. PPACD is a distinctive form of primary choroidal dystrophy affecting the central macula, and all forms of these dystrophies exhibit distinctive atrophy patterns. Here, we present a case of PPACD and emphasize the importance of this disease group by providing a brief summary of primary choroidal dystrophies affecting the central macula.

Keywords: Central macula, choroidal dystrophies, fluorescein angiography, posterior polar annular choroidal dystrophy

INTRODUCTION

Posterior polar annular choroidal dystrophy (PPACD), a form of primary choroidal dystrophy affecting the central macula, is a rare ocular disease characterized by atrophy of the retinal pigment epithelium (RPE) and choriocapillaris encircling the optic nerve head and retinal vascular arcades.[1] Since Yanuzzi initially described the term PPACD in 2010,[2] only a limited number of cases have been reported because of the rarity of the condition. The objective of this study was to present a case of PPACD and discuss other forms of primary choroidal dystrophies that affect the central macula, with a comparison to our case.

CASE

A 56-year-old female patient who was referred to the retina clinic underwent evaluation. The patient denied acute vision loss and had no significant medical history. There was no family history of hereditary eye disease. Upon visual acuity examination, the patient's best-corrected visual acuity was 20/32 in both eyes. With Goldmann applanation, the intraocular pressure (IOP) was 13 mmHg on the right and 17 mmHg on the left. Facial symmetry, external face, and ocular alignment were normal on examination. The anterior segment slit-lamp examination was unremarkable in both eyes. Dilated fundus examination of each eye revealed chorioretinal atrophy surrounding the optic disc extending along the temporal vascular arcade, forming an annular pattern with preservation of the fovea. Atrophic appearance was more extensive in the left eye than in the right eye. Sporadic pigment clusters were observed in both eyes. No arteriolar attenuation or bone spicule-like pigmentary changes were observed. Examination of the optic nerve head and peripheral fundus revealed no additional pathology (Figure A, B).

Fluorescein angiography showed that large choroidal vessels could be visualised along the vascular arcades in the right eye and around the macula in the left eye in a concentric pattern due to the atrophy of the RPE and the choriocapillaris. Additionally, late-phase staining in the atrophic region surrounding the optic nerve head was observed in both the eyes (Figure C, D). Fundus autofluorescence imaging revealed well-defined hypoautofluorescence in an annular pattern around the fovea and optic disc, corresponding to the regions of chorioretinal atrophy. On the basis of these findings, a diagnosis of PPACD was established.

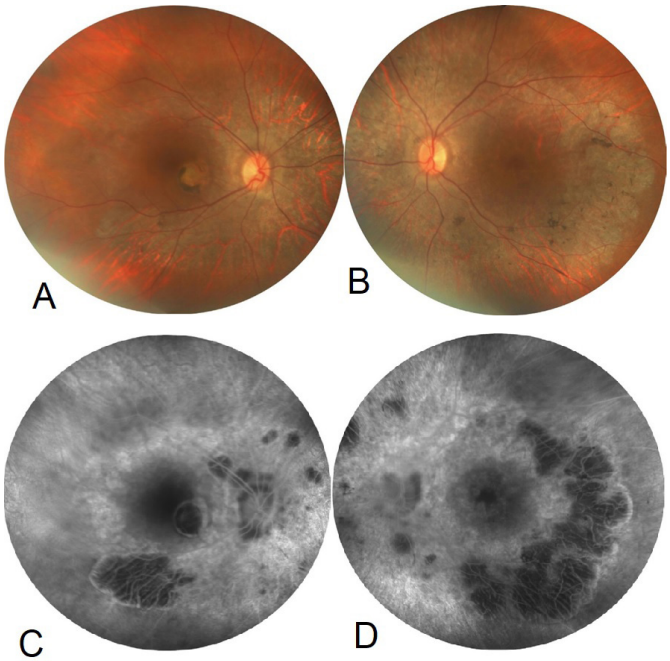

Figure. (A) Color fundus photographs of the right and (B) Left eyes demonstrate bilateral peripapillary atrophy extending along the temporal arcades in an annular pattern. (C) Fluorescein angiography of the right and (D) Left eyes reveals extensive retinal pigment epithelium and choriocapillaris atrophy, allowing the visualization of large choroidal vessels

DISCUSSION

Primary choroidal dystrophies affecting the central macula constitute a discrete group of diseases that exhibit diverse patterns of atrophy, affecting both the retinal pigment epithelium (RPE) and choriocapillaris. The following conditions are considered primary choroidal dystrophies affecting the central macula: central areolar choroidal dystrophy, posterior polar central choroidal dystrophy, posterior polar annular dystrophy, posterior polar hemispheric dystrophy, and central and peripheral annular choroidal dystrophy.[1]

Central areolar choroidal dystrophy (CACD) usually manifests with localized parafoveal pigmentary alterations at the initial stage of the disease, which then progress into a bilateral, symmetrical, atrophic region at the center of the macula.[3] The atrophic region is well-defined and delineated from the surrounding normal retina by distinct borders. With atrophy of the RPE and choriocapillaris, the larger choroidal vessels become apparent. While instances of autosomal recessive transmission have been reported, autosomal dominant inheritance resulting from mutations in the peripherin-2 gene is the prevailing pattern in the majority of cases. Differential diagnosis should be made, especially with atrophic age-related macular degeneration.[3] In contrast to PPACD, there are no areas of atrophy along the vascular arcades or surrounding the optic disc.

In posterior polar central choroidal dystrophy, atrophic anomalies affect the posterior fundus within the vascular arcades and occasionally extend to the region surrounding the optic nerve. Initially, the disease may present as a localized degenerative process, later progressing to involve the entire posterior pole.[4] In PPACD, the fovea is typically preserved, as observed in our case.

In posterior polar hemispheric dystrophy, the atrophic alteration affects half of the posterior pole, extending from the juxtafoveal area to the vascular arcade.[4] In PPACD, a ring-like atrophy is observed along the vascular arcades. With the case report documenting posterior polar annular and hemispheric dystrophy in the same patient, there is a proposition that these two conditions might constitute a single entity.[5]

Central and peripheral annular choroidal dystrophy are distinguished by bilateral symmetrical atrophy at the posterior pole, similar to that observed in central choroidal dystrophy. This is accompanied by a sizable ring-like atrophic region and pigmentary alterations on the peripheral fundus.[4] To date, no abnormalities have been reported in the peripheral fundus in patients with PPACD.[6]

While bilateral involvement was observed in all cases of PPACD, asymmetricity was also a common phenomenon.[1] In our case, the atrophic area was more prevalent in the left eye.

Fluorescein angiography typically reveals a window defect in the atrophic region.[6] However, in a study by Forte et al.,[7] hyperfluorescence was observed in atrophic areas. In contrast, our patient showed diffuse hypofluorescence with large choroidal vessels that were easily visible. This suggests that the choriocapillaris is also extensively affected over time by RPE. Furthermore, Narayanan et al.[5] demonstrated a notable reduction in retinal vasculature in the deep capillary plexus as well as loss of the choriocapillaris in the affected regions. In our case, the presence of diffuse RPE and choriocapillaris atrophy with [l]arge choroidal vessels becoming visible on fluorescein angiography may be indicative of late-stage PPACD. Indeed, it has been reported that the area of atrophy spreads and enlarges insidiously in long term follow-up, and cystoid macular edema and retinal vascularization may develop.[7-9] The utilization of multimodal imaging modalities, including fundus autofluorescence, optical coherence tomography, electroretinogram (ERG) and automated perimetry, is also a valuable approach in the diagnosis of PPACD.[7]

Short-wavelength fundus autofluorescence typically shows peripapillary hypoautofluorescence extending to the vascular arcades in an annular pattern, consistent with the areas of the chorioretinal atrophy. On occasion, a perifoveal hyperautofluorescent border may be observed, which indicates the transition between the healthy and atrophic zones.[6] The hyperautofluorescent border indicates an increase in outer segment phagocytosis and lipofuscin accumulation in RPE. This phenomenon is believed to be the initial stage of atrophy, which will subsequently spread to this region over time.[7]

Two case reports have documented a reduction in amplitude of the scotopic and photopic a-waves and b-waves, as well as a decreased 30Hz flicker response and oscillatory potentials with delayed implicit time in the full-field ERG analysis.[1,5] Sone et al.[8] demonstrated a mild reduction in a-wave amplitudes in scotopic and combined rod-cone responses, as well as a slight decrease in cone and 30-Hz flicker responses. Although the chorioretinal atrophy is confined to the posterior pole in PPACD, the reduction in the amplitude of photopic and scotopic a-wave is remarkable in suggesting diffuse photoreceptor dysfunction. The precise genetic etiology of PPACD remains unclear. However, the presence of diffused depressed responses in the full-field ERG, in conjunction with the clinical presentation and symptoms

such as night blindness, suggests that PPACD may share a similar genetic basis to inherited retinal dystrophies.[6] In contrast to the results reported in the literature, Valle et al.[10] reported a decrease in cone response and a normal rod response in the ERG analysis.

Perimetry showed ring scotoma in two previous reported cases.[5,8] In the case report published by Sone et al.,[8] kinetic perimetry revealed that the ring scotoma was consistent with areas of chorioretinal atrophy and that there was no concentric constriction of the visual field. In a study conducted by Gala et al.,[1] static automated perimetry demonstrated generalized depressed points, particularly in the pericentral area. Furthermore, an enlarged blind spot due to peripapillary atrophy has been documented.[6]

Although no structural or vascular abnormalities can be detected in the fovea with current imaging methods, and the abnormality is mostly around the vascular arcades, the underlying cause of visual acuity loss in patients has not yet been clearly elucidated. A case report by Narayanan et al.[5] demonstrated diffuse functional dysfunction in photoreceptors on the ERG. Another case, as reported by Valle et al.,[10] demonstrated cone dysfunction on ERG. Furthermore, a case study utilising adaptive optics imaging observed the loss of cone photoreceptors in the fovea.[7] When all the evidence is taken together, it may be suggested that the involvement of foveal cone photoreceptors in PPACD cases may result in a deterioration of visual quality or even a decline in visual acuity, as observed in our case. Further investigations are required to elucidate the cause of visual impairment while the fovea remains spared.

CONCLUSION

Primary choroidal dystrophies that affect the central macula constitute a discrete group of diseases that exhibit diverse patterns of chorioretinal atrophy. PPACD is a rare disease that is considered one of the primary choroidal dystrophies affecting the central macula and typically preserves the fovea.

ETHICAL DECLARATIONS

Informed Consent

The patient signed and free and informed consent form.

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Financial Disclosure

The authors declared that this study has received no financial support.

Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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