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Case report

Unilateral pigmented paravenous retinochoroidal atrophy with retinitis pigmentosa in the contralateral eye: A case report

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ABSTRACT

Purpose: We describe a sporadic case of unilateral pigmented paravenous retinochoroidal atrophy (PPRCA) with retinitis pigmentosa (RP) in the contralateral eye.

Observations: a 24-year-old female aware of the narrowing of visual field was examined at our hospital. Funduscopic examination revealed left eye showing retinochroidal atrophy along the retinal veins with pigment accumulation while right eye showing peripheral diffuse retinal pigmented epithelium atrophy with bone spicule pigmentation. Fundus autofluorescence, electroretinogram, visual field test and optic coherent tomography were also performed and obtained results were compatible with funduscopic observation.

Conclusions and importance: Simultaneous manifestation of PPRCA and RP observed in this case is rare and supports a shared genetic basis between the two diseases. Further genetic investigations are needed to elucidate the etiology and to properly manage PPRCA.

1. Introduction

Pigmented paravenous retinochoroidal atrophy (PPRCA) is a rare type of retinochoroidal atrophy with pigment accumulations lesion of which is observed characteristically along retinal veins in a bilateral manner.[1] Although PPRCA patients are usually asymptomatic and show no or subtle progression,[2,3] effective treatment has not been identified.

The disease etiology remains unclear. The majority of PPRCA patients are sporadic, whereas recent studies on minor familial cases indicate the hereditary nature.[4–6] In addition, an inflammatory etiology has been proposed. Previous reports demonstrated that PPRCA follows various inflammatory causes, including congenital syphilis, measles, rubeola and uveitis.[7–10]

In the present study, we report a unilateral case of PPRCA with retinitis pigmentosa (RP) observed in the fellow eye. Our findings support shared genetics between PPRCA and RP.

2. Case report

In 2014, a 24-year-old female was referred for detailed fundus examination. She has been aware of narrowed visual field for the right eye since teenager, while she had no complaints on the left eye. There was no remarkable systemic, ocular, medical or familial history. Best corrected visual acuity in decimal fraction at the first visit was 1.2 in both eyes with refraction of −0.75 OD and −3.75 OS, respectively. The anterior segments and media were normal.

Funduscopic examination together with fundus autofluorescence (FAF) imaging technique revealed that the right and left eye each showed different abnormalities. The left eye had peripapillary retinochroidal atrophy that extends along the retinal veins with bone spicule pigment accumulation distributed mainly at distal parts of the atrophic areas, leaving the macula and the optic disc intact. The right eye had an unaffected macula surrounded by peripheral retinal pigmented epithelium (RPE) atrophy with diffuse bone spicule pigmentation. Fluorescein angiography showed no signs of inflammation in both eyes. We observed arcs of increased autofluorescence surrounding the area of RPE atrophy in the left eye, while parafoveal ring-shaped area of increased autofluorescence in the right eye (Fig. 1). Optical coherence tomography (OCT) demonstrated that ellipsoid zone was absent in the arcuate

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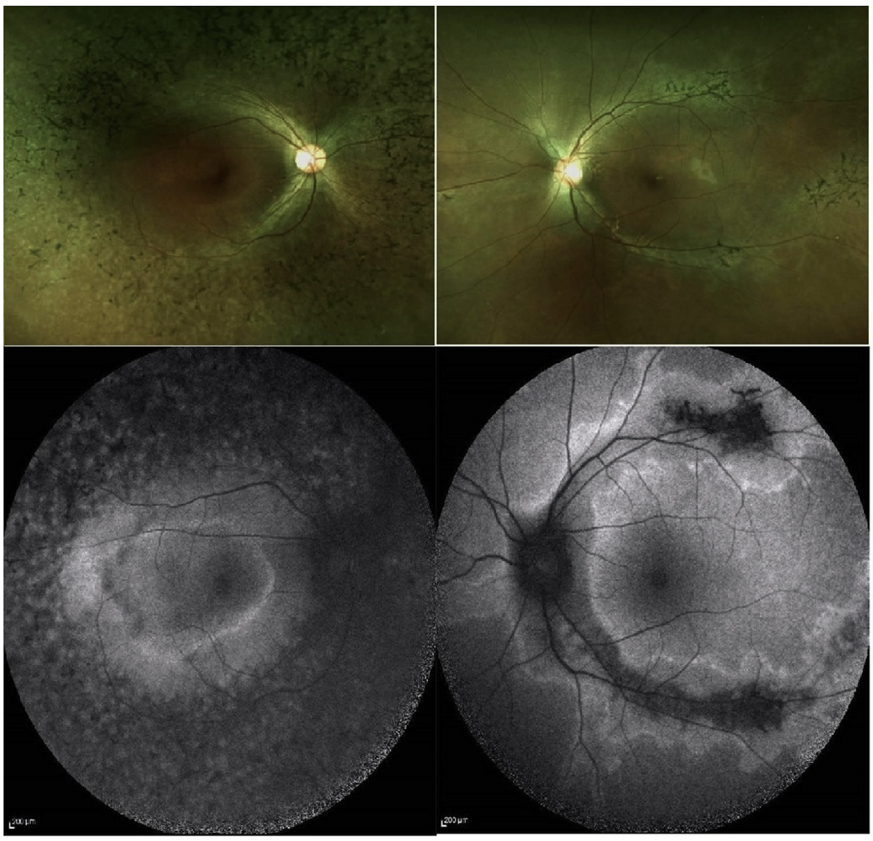

Fig. 1. Fundus photography together with autofluorescence. (Top Left) The right eye showed peripheral retinal pigmented epithelium atrophy with diffuse bone spicule pigmentation, (Top Right) while the left eye had paravenous retinochroidal atrophy. (Bottom Left) We observed parafoveal ring-shaped increased autofluorescence in the right eye, (Bottom Right) while arcs of increased autofluorescence surrounding the area of retinal pigment epithelium atrophy in the left eye.

area or parafoveal ring-shaped area with increased FAF in the left and right eye, respectively (Fig. 2). Humphrey field analyzer (HFA) revealed an enlargement of physiologic blind spot and paracentral arcuate scotoma with the left eye and a concentric contraction of visual field with the right eye (Fig. 3).

Electroretinogram (ERG) showed a clear difference between both eyes. The right eye demonstrated non-recordable pattern from all of flash, rod, cone and flicker ERGs, which is compatible with RP. The left eye showed reduced amplitudes in the flash, rod, cone and flicker ERGs with, in particular, reduced b:a ratio in the flash ERG (Fig. 4).

All results of general physical examination and serological tests were negative, indicating no signs of inflammatory disorders such as uveitis.

McKay et al. previously identified V162 M (Val162Met) mutation within exon 2 of CRB1 gene.[11] To investigate genomic sequence, genomic DNA was extracted from blood sample of the patient under consent. However, DNA sequence analysis did not find V162 M mutation (data not shown).

The patient has received no treatment and been followed up with funduscopic examination, FAF, ERG, HFA and OCT. No clinical progression has been observed with both eyes during 3-year follow-up period.

3. Discussion

This case presented a unilateral PPRCA with RP in the contralateral eye. The funduscopic observation showed typical PPRCA phenotype in the left eye, the pigmented region along retinal veins accompanying retinochoroidal atrophy, and RP in the right eye. The results of FAF, HFA and ERG strongly supported our diagnosis.

RP and PPRCA are known to usually manifest in a symmetric manner and regarded as distinct entities, but previous studies indicate they shares genetics. Traversi et al. described a familial case where PPRCA and RP manifested in two siblings and their mother, respectively. They hypothesized that the pigment epithelial damage has been transmitted with heterogenous expression, leading to different clinical phenotype, RP and PPRCA.[12] Recently, Ratra et al. first reported, as the present case, concurrent manifestation of RP in one eye and PPRCA in the other eye found in a single patient.[13] Recent genetic research has found CRB1 (Crumbs 1) gene, the mutations within which are associated with both PPRCA and several types of retinal dystrophies, including retinitis pigmentosa 12, a severe type of RP.[11,14–17] The assumption is reasonable that different mutations within CRB1 gene could cause both PPRCA and RP through different developmental processes. However, in the DNA sequence analysis conducted for this study, V162 M mutation, known to be associated with PPRCA, was not found. Further genetic analysis, including full-length CRB1 gene, is needed to clarify the pathophysiology of PPRCA.

Two possible reasons are considered for concurrent manifestation of the PPRCA and RP in the present case. First, some incidental event might occur that made both eyes manifest distinct conditions within process of eye development. That means in this case the development of PPRCA or RP depended on environmental factors. A second possible reason is that the patient has genetic mosaic and

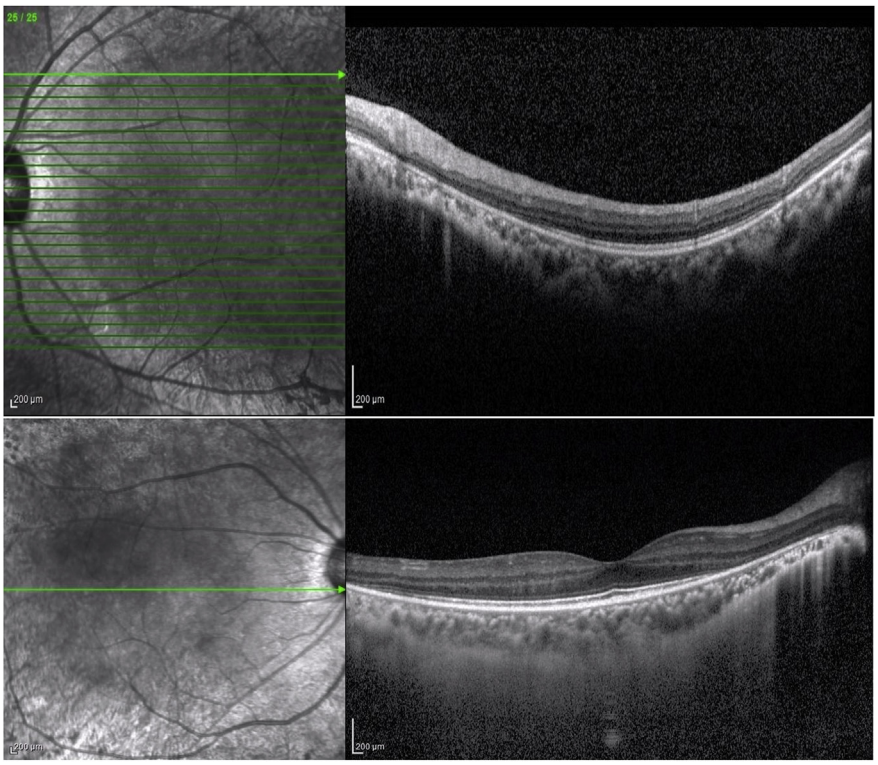

Fig. 2. Optical coherence tomography. (Top) Ellipsoid zone was absent in the arcuate area with increased FAF in the left eye and also (Bottom) in the parafoveal ring-shaped area in the right eye.

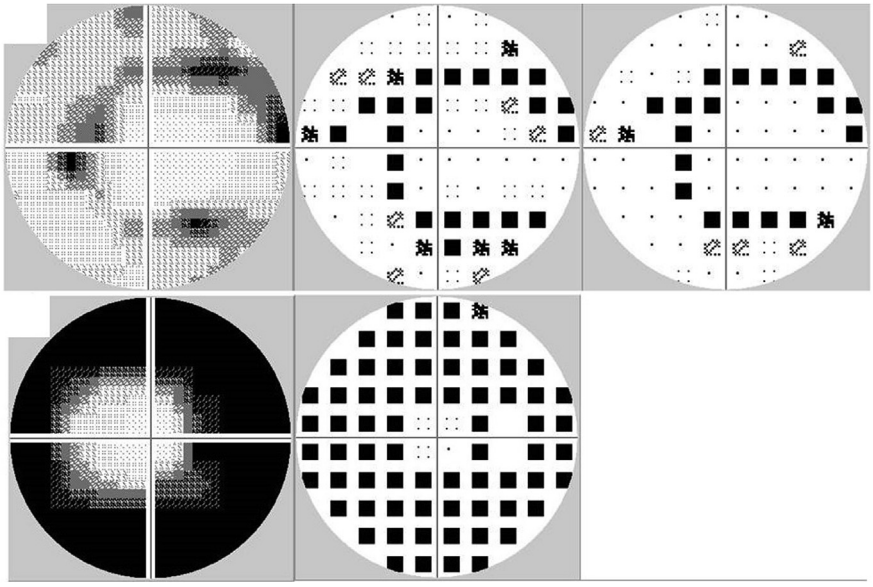

Fig. 3. Humphrey visual field revealed (Top) paracentral arcuate scotoma with an enlargement of Mariotte scotma in the left eye and (Bottom) a concentric contraction of visual field in the right eye.

two distinct genetic types between cell populations determined to become her right eye and left eye led to the two distinct phenotypes, PPRCA and RP.

We should take the case of asymmetric RP manifestation into consideration. Although the observed fundoscopic feature in the left eye was typical to PPRCA, there is a possibility that this gradually progresses to RP in the future, as suggested by the present abnormal ERG series. Some PPRCA patients develop RP after diagnosis or their family member develops retinal degenerative disease, as previously reported.[12]

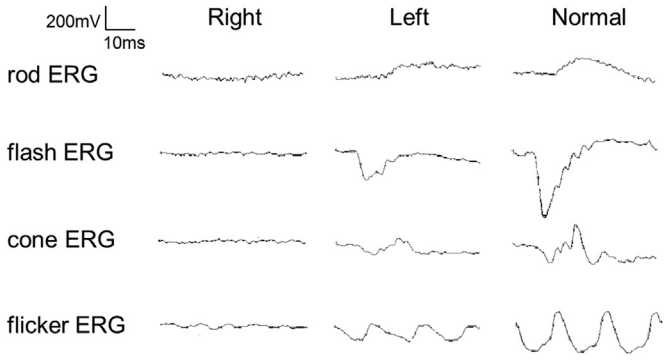

Fig. 4. Electroretinogram (ERG). The right eye demonstrated a non-recordable ERG. The left eye showed reduced amplitudes in the flash, rod, cone and flicker ERG.

4. Conclusions

Simultaneous manifestation of PPRCA and RP in a single patient was observed. The present case supports a shared genetics between the two disorders. Further case accumulation and genetic investigation are needed to elucidate the etiology and proper management of PPRCA.

Informed consent

Consent to publish the case report was not obtained. This report does not contain any personal information that could lead to the identification of the patient.

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Conflict of interest

The following authors have no financial disclosures: Shuichiro Aoki, Tatsuya Inoue, Mari Kusakabe, Masaya Fukushima, Kohdai Kitamoto, Motoshi Yamamoto, Asako Ogawa, Ryo Obata.

Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

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