Bilateral Pigmented Paravenous Retinochoroidal Atrophy With Vasculitis

A case report of a patient with a rare condition.

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Retinitis pigmentosa (RP) is a group of hereditary disorders characterized by progressive functional loss of photoreceptors and retinal pigment epithelium (RPE). Pigmented paravenous retinochoroidal atrophy (PPRCA) is a rare condition in which atrophic areas extend on either side of a vein and follow the vein's course, even when it branches. The cause of this condition is still unknown. Yamaguchi et al[1] reported a 47-year-old Japanese man who had a progressive degeneration of the retina and choroid along the retinal veins associated with uveitis of 2 years' duration. Although steroid therapy cleared the anterior and posterior uveitis in their patient, it had no effect on the inflammatory PPRCA.

Batioglu et al[2] reported a case of PPRCA with ocular inflammation and cystoid macular edema (CME). The association of active inflammation with CME was unique in this case. There have been limited numbers of reports of active inflammation with this disease.[1,2]

Both the above-mentioned reports, however, had no evidence of vasculitis. Herein, we report a case of PPRCA with vasculitis.

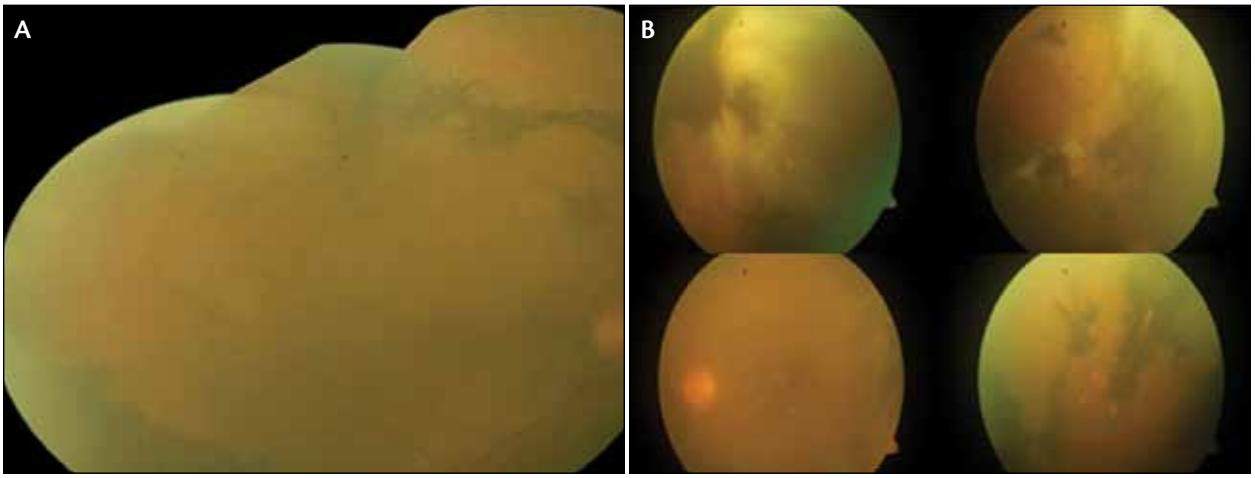

Figure 1. Fundus OD showing pigmentation along the vessels (A). Fundus OS showing pigmentation along the vessels and exudation in the superotemporal quadrant (B).

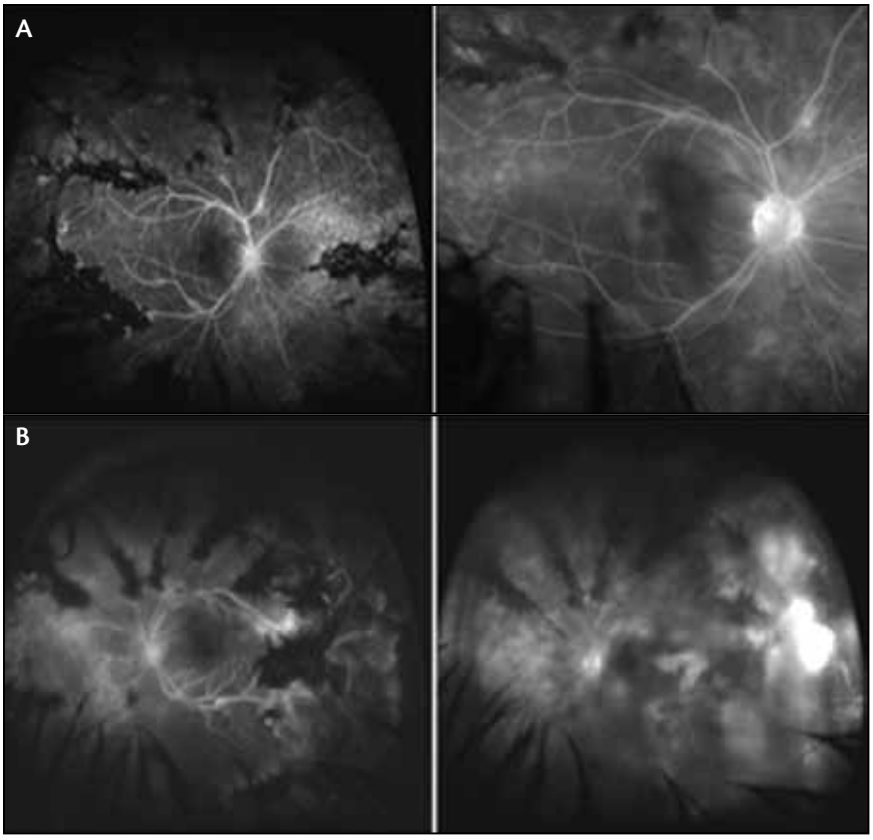

Figure 2. Hypofluorescence OD in late phase corresponding to retinal pigmentation along the vessels (A). Hypofluorescence OS in late phase corresponding to retinal pigmentation along the vessels with leakage in the superotemporal quadrant (B).

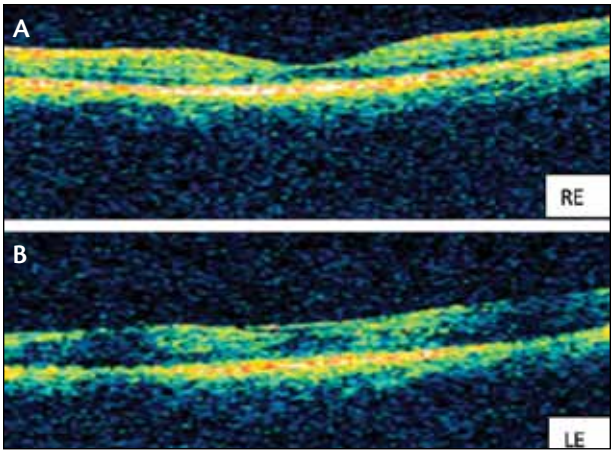

Figure 3. Normal OCT OD (A). OCT OS shows minimal diffuse macular edema with epiretinal membrane (B).

CASE REPORT

A 45-year-old woman presented with complaints of worsening of vision over the past 2 months and poor night vision since childhood. She was a product of a nonconsanguineous marriage.

Ophthalmic examination revealed a distance best corrected visual acuity (BCVA) of 20/30 in the right eye (OD) and 20/240 in the left eye (OS). Slit-lamp examination disclosed inflammation in each eye (OU) with occasional cells OD and 3+ cells OS, bilateral posterior subscapular cataract, and intraocular pressures of 12 mm Hg OD and 14 mm Hg OS. Fundus examination revealed hyperpigmentation along the retinal vessels OU with exudation along the vessels OS more than OD (Figure 1).

Fluorescein angiography showed hypofluorescence corresponding to the retinal pigmentation along the vessels OU. Increased intensity due to leakage suggestive of active vasculitis was noted OS associated with leakage in the macular area corresponding to macular edema (Figure 2).

Optical coherence tomography was normal OD but revealed an epiretinal membrane with diffuse macular edema OS (Figure 3). Electrophysiologic tests were performed showing subnormal A- and B-wave amplitudes in rod response (scotopic electroretinography [ERG]) and maximal combined response and cone response (photopic ERG) OU (Figure 4).

No abnormalities were found on systemic examination. Results of laboratory studies, including a vasculitis workup, were within normal limits. A tuberculin skin testing result was 20 mm, erythrocyte sedimentation rate was 23 mm/hr, and a Quantiferon TB gold test (Qiagen) result was normal. There was no serologic evidence of syphilis or toxoplasmosis.

The patient received antitubercular therapy and systemic steroids under a physician's supervision. Improvement in symptoms was noted at 2 weeks. Subsequent follow-up at 6 weeks showed a distance BCVA of 20/30 OD and 20/120 OS with decrease in signs of vasculitis OS (Figure 5), which was maintained at subsequent follow-ups.

DISCUSSION

The diagnosis of PPRCA is based on the appearance

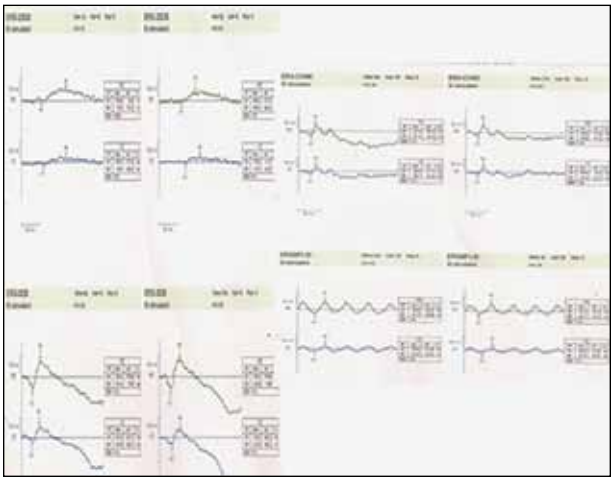

Figure 4. Full-field ERG OU shows subnormal wave forms.

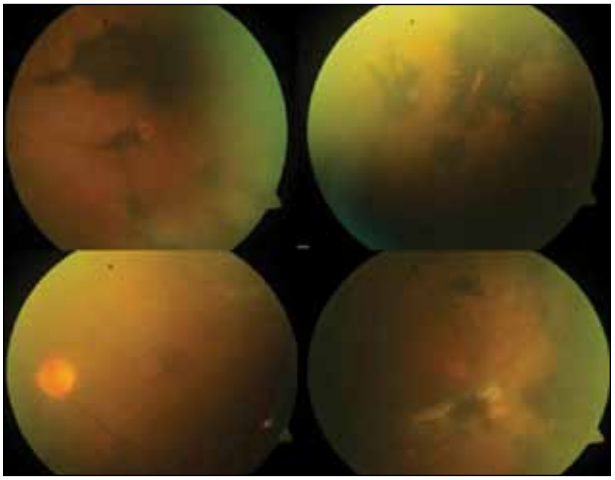

Figure 5. Fundus photo OS shows pigmentation along the vessels and improvement in signs of exudation in the superotemporal quadrant.

of areas of atrophy of the pigment epithelium, and choriocapillaris around the optic disc and along the retinal vessels and is a variant of RP with slow progression.[3]

The etiology of PPRCA is unknown but has an association with CRB1 gene mutation. In a case series,[3] men were more likely to exhibit a severe phenotype, whereas women may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a Val162Met mutation in CRB1, which is likely to affect the structure of the CRB1 protein.[4] Other suggested etiologies include congenital dysgenesis of the RPE, degenerative RPE, dysfunction of the RPE due to active inflammation or infection (tuberculosis, syphilis), and postinflammatory dysfunction.[5]

Kukner et al[6] followed cases of PPRCA for 18 months and found that fundus abnormalities ranged from mild to severe and that retinal function tests indicated that this was a geographic and not a generalized disorder. A vasculitis component was not noted in any of the cases cited above.

Whether vasculitis and PPRCA have a common genetic predisposition requires further investigation.

Brown and Hsiang[4] suggested that the pathologic process in PPRCA is primary rather than secondary to an inflammatory periphlebitis-like condition.

Paravascular pigmentation, which is the hallmark of PPCRA, and signs of vasculitis were findings in the case presented here. A positive tuberculin skin test and signs of inflammation were noted. Systemic steroids and antitubercular treatment resulted in reduction of inflammation leading to visual improvement in the left eye.

The etiology in this case could be inflammatory, infectious, or both. To the best of our knowledge after an extensive literature search, PPCRA with vasculitis has not been previously reported. Although a genetic association with PPRCA is well known, whether vasculitis and PPRCA have a common genetic predisposition requires further investigation. ■

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