Concurrent retinitis pigmentosa and pigmented paravenous retinochoroidal atrophy phenotypes in the same patient

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We report a unique case of a patient with retinitis pigmentosa (RP) phenotype in one eye and pigmented paravenous retinochoroidal atrophy (PPRCA) phenotype in the other eye. We describe in detail the symptoms, clinical findings, and investigations done for a 32-year-old Indian woman. This patient had phenotypical picture resembling typical RP in the right eye, with characteristic symptoms of night blindness and constricted field of vision and a nonrecordable electroretinogram (ERG). The left eye of the same patient revealed typical PPRCA phenotype, with no night blindness, normal field, and normal ERG. RP and PPRCA phenotypes are part of the same spectrum of genetic disorder. However, it is rare to see them coexist in the same patient.

Key words: CRB1 gene mutation, hereditary disorders, night blindness, pigmented paravenous retinochoroidal atrophy, retinal degeneration, retinitis pigmentosa, unilateral retinal degeneration

Retinitis pigmentosa (RP) is a genetic disorder characterized by early retinal degeneration, resulting in difficulty in night vision and progressive peripheral field loss. The symptoms as well as clinical findings are usually bilaterally symmetrical. However, there have been several reports of unilateral RP.[1] Pigmented paravenous retinochoroidal atrophy (PPRCA) is also commonly a bilaterally symmetrical condition. Patients are usually asymptomatic and it is often incidentally detected in a routine examination. There have been reports of marked asymmetry and unilaterality.[2] We report a unique case with RP phenotype in one eye and features suggestive of PPRCA phenotype in the other eye of the same patient.

Case Report

A 32-year-old woman presented to us with the chief complaint of difficulty in night vision in her right eye. She had no visual complaints in the left eye. She had no history of any systemic disease, malignancy, chronic drug intake, or any inflammatory eye disease in the past. There was no family history of night blindness. The best corrected visual acuity was 20/50 in the right eye and 20/30 in the left eye. There was a relative afferent pupillary defect in the right eye. The intraocular pressure was 10 mmHg in the right eye and 20 mmHg in the left eye. Fundus examination showed waxy pallor of the disc, arteriolar attenuation, bony spicule pigmentation, diagnostic of RP, in the right eye [Fig. 1a]. The left eye showed a normal pink-colored disc and no attenuation of arterioles or bony spicules. Pigmented atrophic patches were seen along the vessels [Fig. 1b]. There were no signs of any past inflammation in either eye. Investigations were ordered to confirm these findings. All tests pointed to a diagnosis of RP in the right eye. There was generalized hypoautofluorescence of the retina [Fig. 2a]. Optical coherence tomography of the macula showed loss of the ellipsoid zone and external limiting membrane with retinal thinning [Fig. 2c]. Electroretinogram (ERG) of the right eye showed nonrecordable scotopic and photopic responses [Fig. 3a] and a multifocal ERG showed nonrecordable foveal, parafoveal, and perifoveal ring responses [Fig. 3b]. In addition, there was gross peripheral constriction of the visual field [Fig. 4a]. However, the left eye showed hypoautofluorescence only along the vessels [Fig. 2b]. The OCT was normal with intact ellipsoid zone and external limiting membrane [Fig. 2d]. There were normal scotopic and photopic responses on ERG [Fig. 3a] and normal foveal, parafoveal,

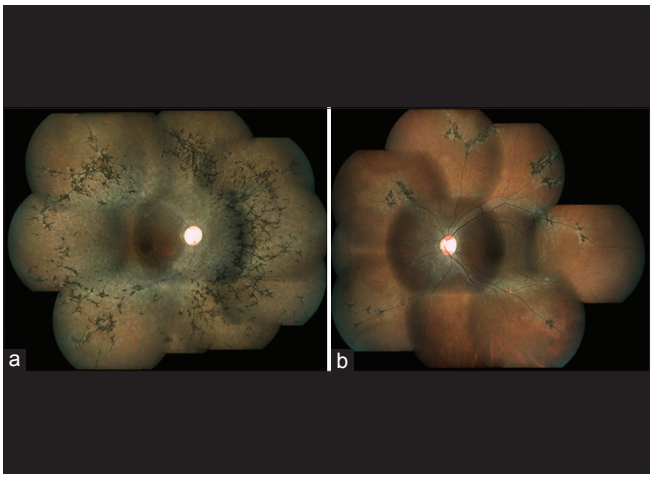

Figure 1: Fundus photograph of the right eye showing waxy pallor of the disc, attenuated arterioles, and bony spicule pigmentation characteristic of retinitis pigmentosa (a). The left eye shows normal disc, normal caliber of arterioles, mild atrophy of the retinal pigment epithelium along the blood vessels, and a few perivascular bony spicule pigments, suggestive of pigmented paravenous retinochoroidal atrophy (b)

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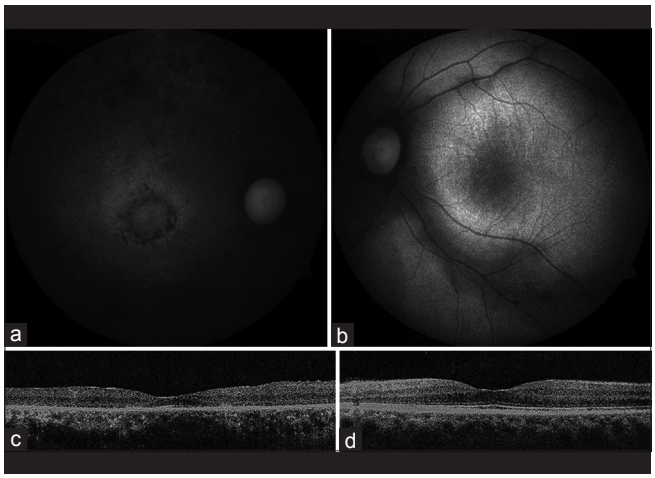

Figure 2: Fundus autofluorescence images of right and left eye. The right eye shows generalized hypoautofluorescence in the peripheral retina and a hyperautofluorescent ring with central hypoautofluorescence at the posterior pole (a). The left eye shows hypoautofluorescence along the vessels only (b). Optical coherence tomography of the right eye showing loss of the ellipsoid zone and external limiting membrane with thinning of the retina (c), optical coherence tomography of the left eye showing intact ellipsoid zone and external limiting membrane (d)

and perifoveal ring responses on multifocal ERG [Fig. 3c]. The visual field was also normal in the left eye [Fig. 4b]. The findings were suggestive of PPRCA in the left eye.

Discussion

Unilateral RP is known to occur where the other eye is mostly seen to be normal. PPRCA is considered to be a separate entity. It was earlier thought to be postinflammatory, but many reports of familial cases make hereditary nature of this condition quite apparent.[2] Interestingly, our patient has both, RP in one eye and PPRCA in the other eye. Despite a thorough literature search using PubMed, Ovid Medline, EMBASE, and Google Scholar, we could not find any report of a similar case. The occurrence of unilateral RP in the mother and bilateral PPRCA in two of her children has been reported, where all these patients showed no changes in ocular findings for 21 years.[3] Various mutations of the CRB1 gene have been implicated in the etiology of PPRCA[4] and also of RP.[5] The PPRCA phenotype has been found to be associated with a Val162Met mutation and three novel mutations of CRB1 gene have been described in RP.[4,5] Therefore, we postulate that our patient may have a hitherto unknown novel mutation of the CRB1 gene, leading to a peculiar phenotypic expression of RP in one eye and PPRCA in the other. We could not confirm the etiology as our patient could not undergo genetic

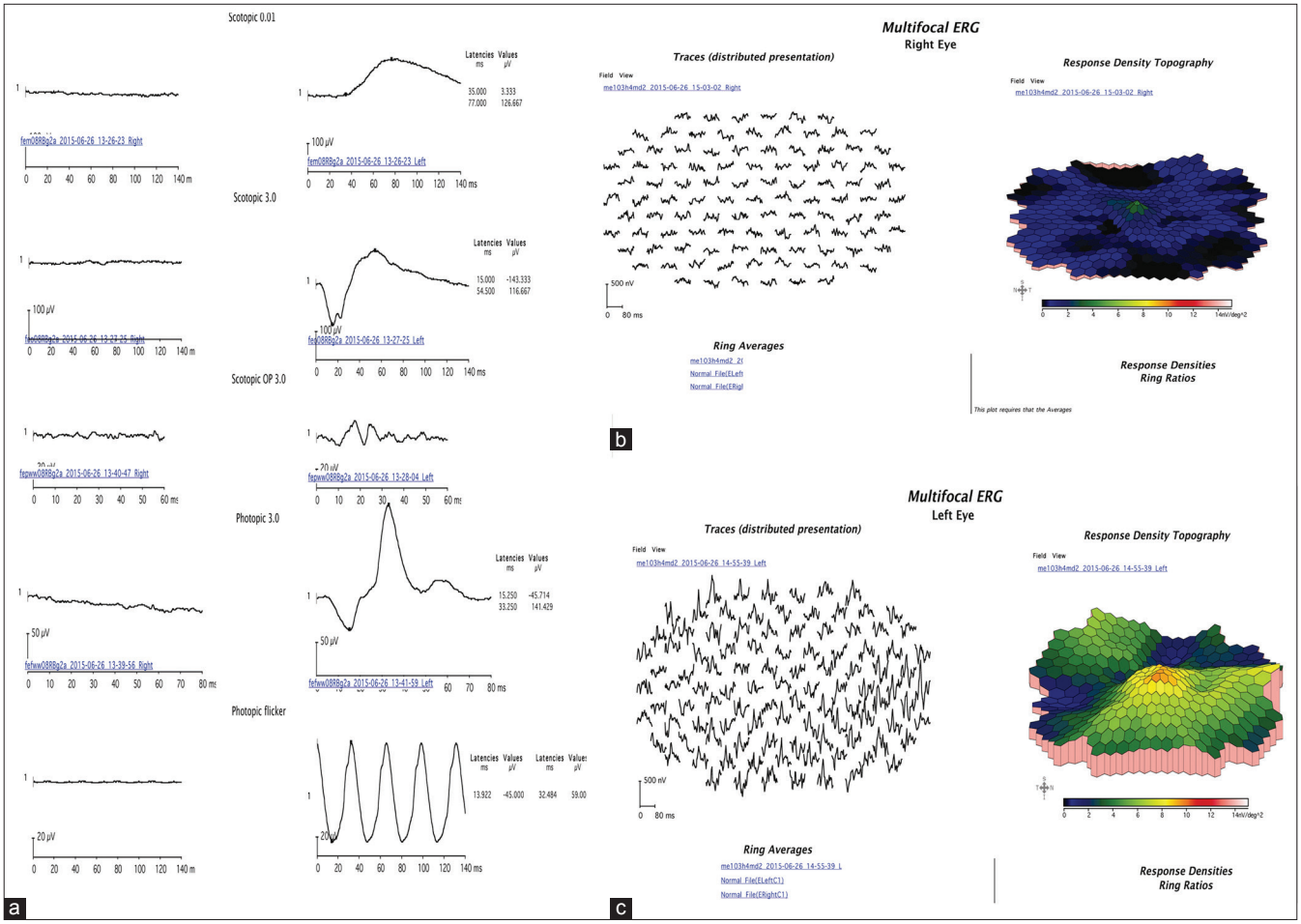

Figure 3: Electroretinogram of the right eye showing nonrecordable scotopic and photopic responses and left eye showing normal scotopic and photopic responses (a). Multifocal electroretinogram of the right eye with nonrecordable foveal, parafoveal, and perifoveal ring responses (b) and left eye showing normal foveal, parafoveal, and perifoveal ring responses in the left eye (c)

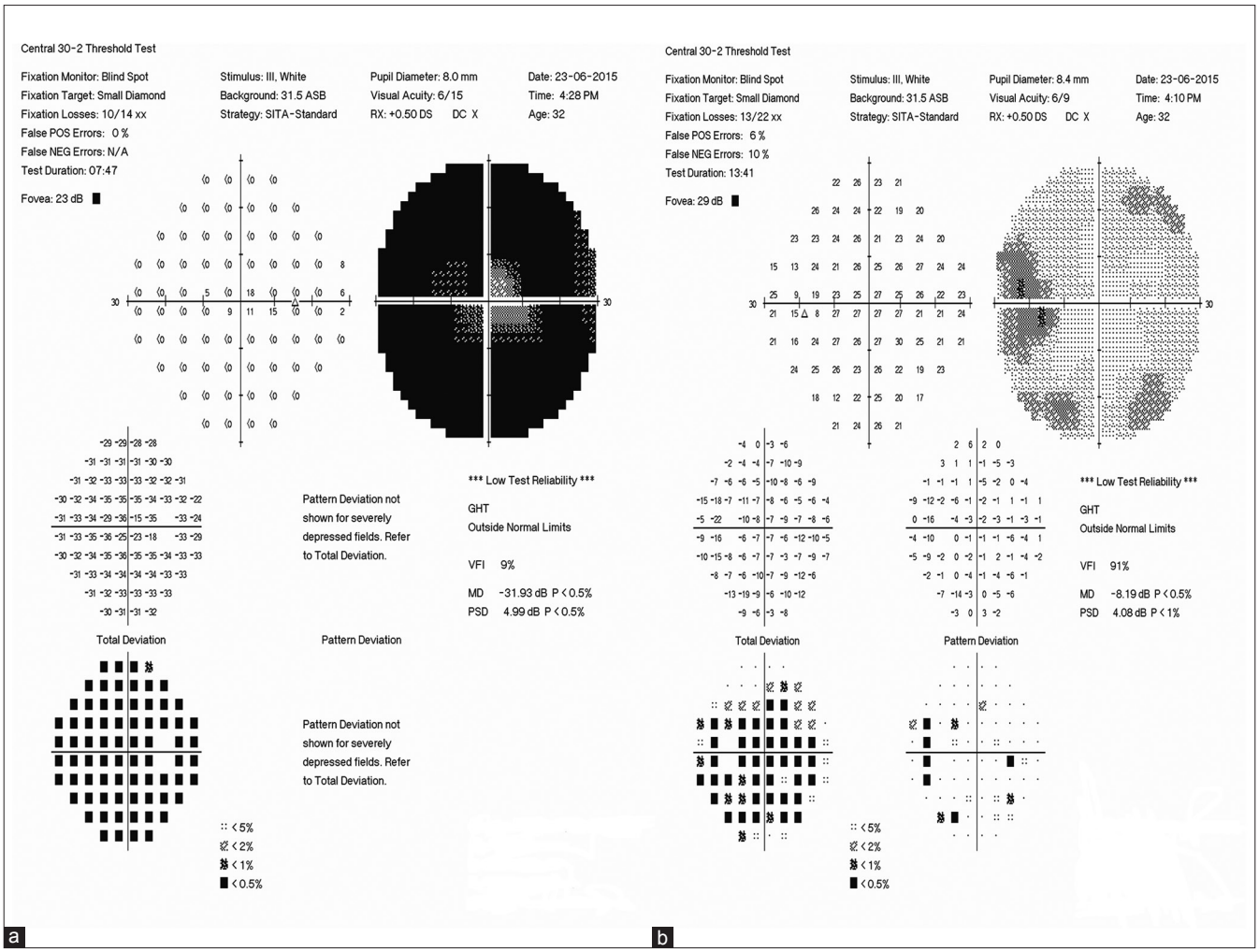

Figure 4: Peripheral constriction of visual fields in the right eye (a) and normal visual fields in the left eye (b)

studies. A long-term follow-up would reveal how the left eye condition progresses, particularly whether RP like degeneration develops in that eye. However, ERG is seen to be abnormal in earliest stages of RP where the retina still appears normal. In our patient, the presence of a normal response on ERG is sufficient to confirm the fact that the left eye is unlikely to develop RP.

In summary, we present an interesting case of two different phenotypes in the same patient that of RP in one eye and PPRCA in the other eye. To the best of our knowledge, this is the first such case to be reported.

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Conflicts of interest

There are no conflicts of interest.

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