

Case Report

A Vision-Saving Straw in a Retinitis Pigmentosa Patient

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Keywords

Retinitis pigmentosa · Cone cell · Blood flow · Cilioretinal artery

Abstract

We report a case of binocular retinitis pigmentosa (RP) with completely different visual acuity between 2 eyes, which may be contributed by the presence of cilioretinal arteries (CRAs) in 1 eye. A 66-year-old female patient complained of blurred vision after binocular cataract surgeries. Examinations revealed her right eye had 20/25 central visual acuity, but the fellow eye only had light perception. Subsequent fundus photography of both eyes firmed the diagnosis of binocular RP. However, there were some significant differences in retinal vessels, which were attenuated in her left eye in contrast to several spared retinal arterioles in the right eye. Optical coherence tomography angiography showed that the spared vessels might be CRAs. Our case provides an evidence that macular blood flow may contribute to the survival of cone cells in RP.

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Introduction

Retinitis pigmentosa (RP) is an inherited retinal disease that is characterized by progressive visual loss, starting from night blindness and leading to impaired visual fields and eventually to complete blindness [1]. It is widely accepted that RP is mainly caused by gene mutations, which result in rod photoreceptor degeneration. However, factors affecting RP progression have not been fully understood yet. A number of recent studies investigated the relationship between ocular hemodynamics and RP, which is vital to understand the pathology of this disease. It has been reported that ocular blood flow (BF) in RP patients is reduced. However, the role of vascular dysfunction in photoreceptor degeneration is not clear, especially in cone cell loss [2].

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In this case, a woman suffering from bilateral RP for about 11 years, who had received cataract surgery on both eyes 2 years ago, showed significantly different best-corrected visual acuity of both eyes. A disparity of vascular structure existed within her both eyes, suggesting a possible reason for the situation of her best-corrected visual acuity. To the best of our knowledge, this is the first report of the vision-retaining effect of arterioles in RP.

Case Report

A 66-year-old female patient came to our service in August 2019, complaining of blurred vision at night for about 11 years. She had experienced binocular cataract surgery successively 2 years ago. Vision examination revealed 20/25 in the right eye (OD) and light perception in the left eye (OS). The fundus photography of her OS revealed characteristic RP fundoscopic findings, which consisted of attenuated retinal arterioles, a waxy pale optic disk, and bone spicule pigmentation. The OD fundus examination indicated similar ghost retinal vessels and retinal pigment clumping; however, the difference was that several spared retinal arterioles which supplied the posterior pole of the retina, and a yellow-white optic disk were observed (shown in Fig. 1). Optical coherence tomography (OCT) examination of the OD indicated a relatively normal thickness of each layer in the macular region and the full thickness of macular lutea was 227 μm , yet the atrophic retinal neurosensory layer and retinal pigment epithelium were detected in the peripheral area. In addition, epiretinal membrane existed. Nevertheless, the OCT image of the fellow eye showed atrophy of the entire retina, and the full thickness of macular lutea was only 114 μm (shown in Fig. 1). To further identify the spared vessels in the OD, we performed an examination of OCT angiography (OCTA), which revealed that the spared vessels might come from the posterior ciliary artery system but not the central retinal artery. But in the fellow eye, the vascular structure is normal (shown in Fig. 2). The visual field measurement exhibited a tubular visual field in the OD eye as well as full blindness in the contralateral eye (shown in Fig. 3).

The patient was diagnosed with binocular advanced RP based on the examination results above. Considering her stage of RP and available therapies, she was eventually treated with neurotrophic agents, by which we hoped to slow the disease progression.

Discussion

We reported a rare case of advanced RP with entirely different visual acuity between 2 eyes, which may be caused by the presence of cilioretinal arteries (CRAs). CRAs are reported to be present in 32% of the eyes and the prevalence of RP all over the world is estimated to be about 1 in 4,000 [3,4]. To our knowledge, there is no similar report of the vision-retaining effect of CRAs in RP.

The primary cause of RP is a variety of different mutations in genes most of which are exclusively expressed in rods. Despite different biological dysfunctions that diverse mutated genes result in, the ultimate common response is the apoptosis of rod cells. It was also assumed that rod cell death in RP contributed to high levels of oxygen in the retina because rods, which are packed with mitochondria and highly metabolically active, compose 95% of the cells in the outer nuclear layer [5]. Yu et al. [6] used a rat model of RP to demonstrate that mean oxygen tension in the outer retina was substantially higher than normal when rods were eliminated and then high levels of oxygen spread into the inner retina. When oxygen levels are high in the inner retina, retinal vessels constrict to reduce BF and oxygen delivery, which

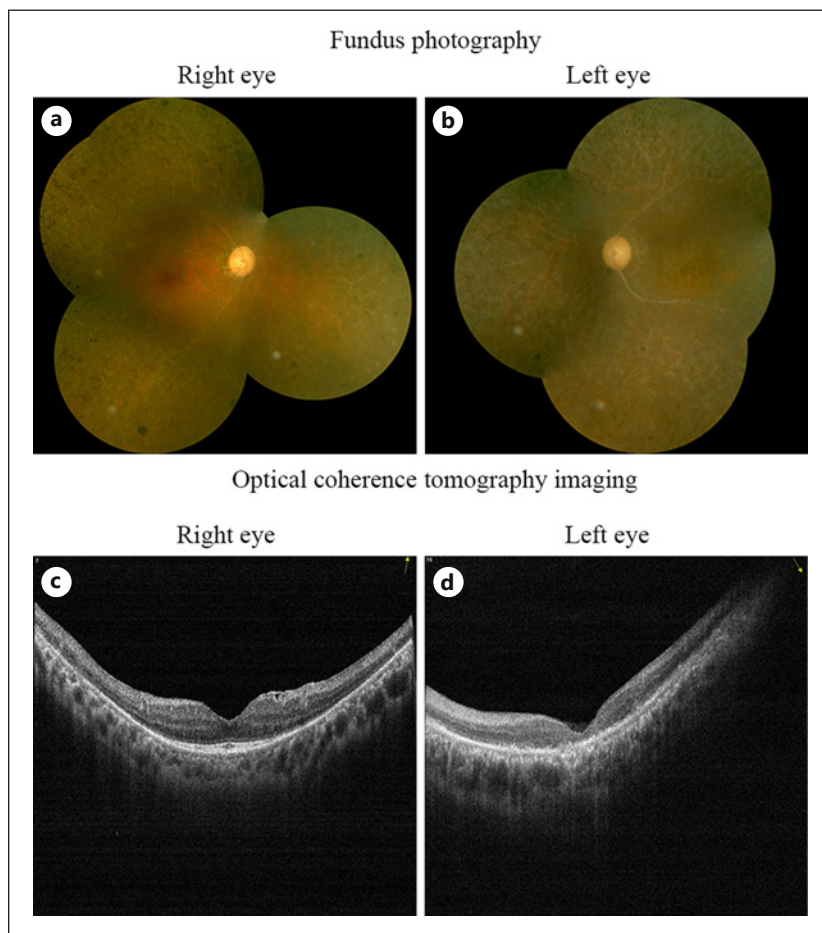


Fig. 1. Fundus examination by fundus photography (a, b) and OCT (c, d). Spared CRAs were observed in the right eye, which may have contributed to the relatively normal thickness of each layer in the macular region in the OCT image. CRA, cilioretinal artery; OCT, optical coherence tomography.

is called autoregulation [5]. However, choroidal BF regulation is distinctly different from the regulation of retinal BF, which is only partly autoregulated [7]. CRAs usually arise from the peripapillary choroid or directly from one of the short posterior ciliary arteries [3]. This reason may explain why CRAs were not attenuated.

Decreased macular BF has been associated with impaired cone cells and central visual function in RP patients. Falsini et al. [8] found that subfoveal choroidal BF measured by laser Doppler flowmetry was reduced in patients with RP and correlated with central cone function assessed by focal electroretinograms. Murakami et al. [9] using laser speckle flowgraphy demonstrated that decreased macular BF was associated with the reduction in macular visual sensitivity in patients with RP and hypothesized that impaired macular BF in RP may contribute to cone cell death. Additionally, another study using OCTA found that parafoveal retinal vascular density was correlated with central visual function and decreased at an earlier stage of RP, suggesting that macular vascular changes may occur concomitantly or followed by cone cell loss [10]. These findings showed that vascular abnormalities in RP were related to decreased central vision and may contribute to cone cell death.

In other fundus diseases, the protective effects of CRAs have already been reported. In permanent nonarteritic central retinal artery occlusion, the presence of CRAs could have a marked influence on the visual outcome and retinal circulation [3]. In another clinical study,

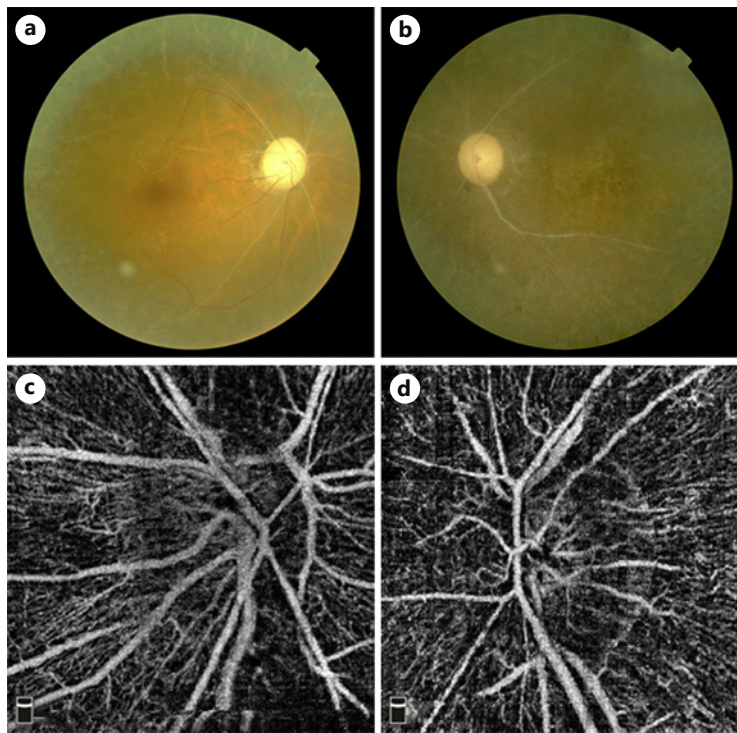


Fig. 2. The fundus photography (a, b) and OCTA image (c, d) of the right eye and the left eye, respectively. OCTA, optical coherence tomography angiography.

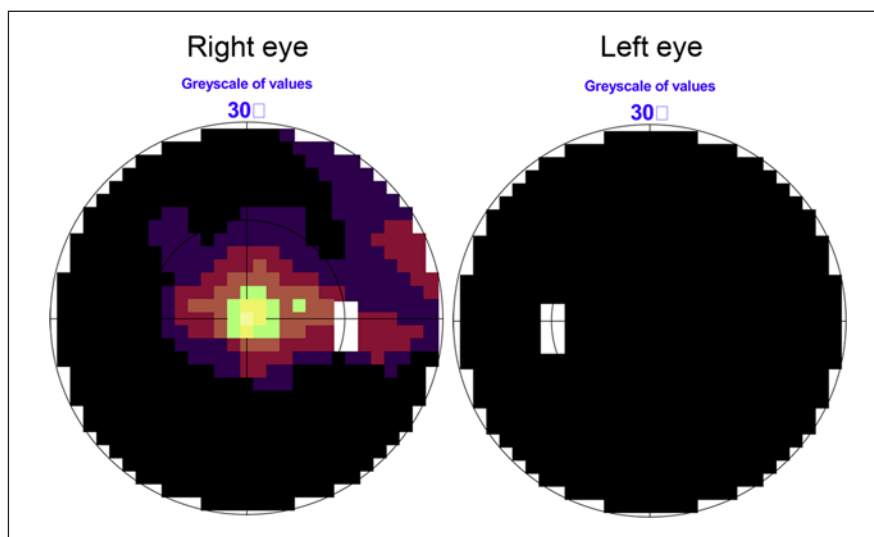


Fig. 3. Visual field examination. The tubular visual field was detected in the right eye, while the central visual field was lost in the left eye.

it was suggested that a CRA could provide more circulation and contribute to retaining central field and visual acuity in advanced open-angle glaucoma [11]. In addition, the finding by Snyder et al. [12] indicated that CRAs were associated with a lower risk of developing choroidal neovascularization. From the findings mentioned above, we suggest that accessory blood supply by CRAs can increase retinal circulation and change the retinal environmental factors.

In this case, the eye with CRAs had significantly better visual acuity than that of the contralateral eye, which could support our theory that CRAs contribute to the survival of cone cells and remaining central vision by providing more macular circulation. Actually, a study showed that macular supply was observed in 76.05% of temporal CRAs [13]. In line with these findings, a clinical study also found that unoprostone increased the macular BF and preserved central visual function in patients with RP [14]. While the previously discussed findings indicate a potential relationship between BF and cone cells, it is important to note that there is a strong evidence that oxidative stress is the cause of cone cell death in RP [5]. The underlying mechanism of BF protection against cone cell death should be further investigated.

RP is an untreatable condition at present. In recent years, gene therapy and cell-based therapies have been areas of active clinical investigation. Although therapeutic attempts have been made, their clinical efficacy has not been clearly proven. Considering the patient's severe ocular state and limitation of available and effective therapies of RP now, she was eventually treated with neurotrophic agents, which have been shown to be effective in protecting photoreceptors in some clinical trials [15].

However, there are some limitations. It would be better to get the patient's fundus fluorescein angiography result, ERG result, and genetic testing result. Unfortunately, the patient refused further examination so we could not fully ascertain the origin of the existing vessels. But according to the OCTA results and the partly autoregulated feature of CRAs, we suspect that the spared vessels may be CRAs.

In summary, our case supports the theory that the existence of arterioles could exert protective effects on RP by modifying the retinal blood supply. Further studies should be conducted to reveal the underlying mechanism of this condition.

Statement of Ethics

The authors have no ethical conflicts to disclose. The patient has given permission to publish this manuscript.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Lijun Wang and Peirong Lu designed the study; Lijun Wang, Jianqing Li, and Peirong Lu participated in the acquisition of data. Lijun Wang, Jianqing Li, Chi Ren, and Peirong Lu analyzed and interpreted the data and wrote the paper. All the authors read and approved the final manuscript.

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