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Peripapillary Retinitis Pigmentosa Complicated with Bilateral Open-Angle Glaucoma: a Case Report

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Abstract: Although the associations between retinitis pigmentosa (RP) with cataract or chronic angle closure glaucoma have previously been widely recognized, the case of cataract and open-angle glaucoma concurrence in peripapillary RP is rarely reported before. Case presentation: A 65-year-old man complained with progressive visual loss in the right eye for one year. The intraocular pressure (IOP) was 44mmHg in the right eye and 27mmHg in the left eye. Ultrasound biomicroscope (UBM) showed the anterior chamber angle was narrow. Optic disc was expanded in the right eye and 0.6 in the left eye. Fundus revealed pigmentation in posterior polar annularly around the macula, waxy pallor of the disc, attenuated arterioles. The two complications-cataract and optic nerve lesion-may simultaneously lead to progressive visual loss. In RP, open-angle glaucoma may be caused by trabecular meshwork inflammation.

1. Introduction

Retinitis pigmentosa (RP) represents a complex and diverse collection of hereditary retinal degenerative disorders, which are characterized by a gradual and progressive decline in the number and functionality of photoreceptor cells within the retina. This decline leads to a range of debilitating symptoms, including night blindness, which severely hampers the ability to see in low-light conditions, and a narrowing of the visual field, which can significantly restrict a person's overall vision. Notably, there exists a well-documented correlation between RP and an increased susceptibility to developing glaucoma, a serious eye condition that can lead to irreversible vision loss[1]. Existing research predominantly associates RP with mechanisms related to angle-closure glaucoma[2], a type of glaucoma that arises from specific alterations in the anterior segment of the eye, including the thickening of the lens and the formation of shallow anterior chambers. In contrast, primary open-angle glaucoma (POAG) is defined by a progressive form of optic nerve damage that is typically linked to dysfunction of the trabecular meshwork, occurring within an open-angle configuration and remaining largely independent of the underlying pathophysiology of RP. Although POAG is widely prevalent in the general population, its occurrence alongside RP is particularly rare, especially in the unusual peripapillary variant, where pigmentary alterations are localized around the optic nerve head[3]. This confluence of conditions introduces significant challenges in diagnosis, as the visual field impairments attributed to glaucoma and those resulting from RP often present similarly, complicating the clinical picture and making it difficult for

healthcare professionals to distinguish between the two conditions effectively.

In this report, we present a noteworthy and compelling case of peripapillary retinitis pigmentosa, which is intriguingly coupled with bilateral primary open-angle glaucoma (POAG), effectively illustrating the complex manner in which these two distinct yet interrelated pathologies collaboratively diminish visual performance and overall ocular health. This account carries substantial clinical implications that warrant careful consideration: firstly, it contests the prevailing and dominant notion that primarily links retinitis pigmentosa (RP) with angle-closure glaucoma, thereby highlighting the critical need for a comprehensive assessment of primary open-angle glaucoma within this specific demographic of patients; secondly, it provides valuable insight into essential differential diagnostic methods that are necessary when two optic neuropathies present concurrently, allowing for a more accurate and nuanced understanding of the patient's condition; and lastly, it examines potential pathophysiological interactions that may occur, such as microvascular impairment and the hastened apoptosis of retinal ganglion cells, which could further complicate the clinical picture. The thorough documentation of this distinctive and multifaceted presentation is intended not only to improve diagnostic precision but also to guide therapeutic interventions for analogous intricate cases, ultimately enhancing patient outcomes and advancing our understanding of these complex ocular disorders.

2. Case Presentation

The patient, a 65-year-old male, presented with a concerning complaint of progressive visual loss in his right eye that had been gradually worsening over the course of one year. He denied experiencing nyctalopia, which is the inability to see well in low light conditions. Notably, he had no documented history of malignancy, chronic drug intake, or any inflammatory ocular diseases that could potentially contribute to his visual impairment. In terms of family dynamics, he has one daughter and one son, with the son exhibiting normal vision and a healthy fundus upon examination. The family ocular history was largely unremarkable, suggesting no hereditary predisposition to eye diseases. Upon examination, the best-corrected visual acuity was measured at Hand Moving/20cm in the right eye, indicating severe visual impairment, while the left eye showed a visual acuity of 0.4. The intraocular pressure (IOP) readings were notably elevated, with 44mmHg in the right eye and 27mmHg in the left eye, raising concerns about potential glaucoma. The axial lengths of both eyes were measured, revealing 24.71mm in the right eye and 24.67mm in the left eye, which are within normal limits. Both eyes exhibited phacoscotasmus and posterior subcapsular cataracts (PSC), as illustrated in Figure 1, but there were no other obvious abnormalities detected in the anterior segment of the eyes.Further investigation using ultrasound biomicroscopy (UBM) revealed that the anterior chamber angle was narrow, as depicted in Figure 2. However, gonioscopy indicated that both eyes had open angles, with no pigmentation observed in the trabecular meshwork, which is often a sign of open-angle glaucoma. The fundus examination revealed significant findings, including waxy pallor of the optic disc, attenuated arterioles, and pigmented atrophic patches that were observed in a posterior polar arrangement around the macula. Autofluorescence imaging highlighted areas of retinal pigment epithelium atrophy, presenting a circular pattern of hypoautofluorescence centered on the macula, as shown in Figure 3. The optic disc ratio was expanded in the right eye, measuring 0.6 in the left eye, as illustrated in Figure 3. Optical coherence tomography (OCT) provided further insights, demonstrating that the ellipsoid zone had disappeared and that the retinal pigment epithelium (RPE) was disrupted, as depicted in Figure 4. Additionally, optical coherence tomography angiography (OCTA) revealed a decrease in choroidal vessel density in the region of retinal atrophy, as shown in Figure 5. Fluorescein angiography (FFA) of the left eye exhibited transmitted fluorescence in the early phase, along with blocked fluorescence corresponding to the area of atrophy, while

hyperfluorescence was noted at the edges of the atrophic zone, as illustrated in Figure 6. Finally, electroretinography (ERG) results for both eyes demonstrated a significant reduction in amplitude for both scotopic and photopic responses, as shown in Figure 7, indicating compromised retinal function.Intraocular pressure was successfully regulated to the normal level, which is defined as ranging from 15 to 18 mmHg in the right eye and from 14 to 17 mmHg in the left eye, through the effective application of topical intraocular pressure-lowering medications that were carefully administered to ensure optimal results.

3. Discussion

Retinitis pigmentosa (RP) is a complex and debilitating condition characterized by a progressive degeneration of the retinal tissue, which typically begins in the peripheral regions of the retina and gradually advances toward the posterior pole, leading to a slow but steady decline in visual acuity that can significantly impact an individual's quality of life[4]. Among the various subtypes of RP, sector RP stands out as an uncommon variant, distinguished by its atypical presentation, where the clinical manifestations are notably restricted to one or two quadrants of the retinal fundus, making it a unique case in the spectrum of retinal disorders[5-6]. The diagnosis of sector RP is predominantly based on a thorough clinical assessment, which requires the careful recognition of a specific “pattern” during the examination process, as this pattern is crucial for identifying the condition accurately[7]. Despite extensive research efforts aimed at unraveling the complexities of this condition, the precise pathophysiology of sector RP continues to be poorly understood; however, it is frequently associated with mutations in the rhodopsin gene, a critical component that plays an essential role in the phototransduction cascade, which is vital for converting light into visual signals. In the case of the patient discussed here, the area of the lesion demonstrated bilateral symmetry, presenting as a distinctive ring-like configuration that encircled the optic disc while notably sparing the macular region, which is often crucial for central vision. Upon conducting a fundoscopic examination, we observed attenuated retinal arterioles and dispersed pigmentation clusters that were localized within the posterior pole, indicative of the underlying retinal changes associated with the disease. Additionally, the findings from electroretinography (ERG) revealed a marked decrease in amplitude for both scotopic and photopic responses, underscoring the significant functional deficits that are associated with the progressive retinal degeneration characteristic of this condition.

Cataract formation is well-documented as one of the most prevalent complications associated with retinitis pigmentosa, a degenerative eye disease that affects the retina and can lead to significant visual impairment[8-9]. In this particular case, the patient exhibited bilateral nuclear opacification, characterized by a clouding of the central part of the lens, along with posterior subcapsular cataracts (PSC), which are known for their tendency to develop at the back of the lens and can severely impact vision. Previous literature has indicated that alterations in aqueous humor factors, which play a crucial role in maintaining the health of the eye, significantly contribute to the risk of developing posterior subcapsular cataracts. This provides valuable insights into the potential association between inflammation and the pathogenesis of PSC development in individuals suffering from retinitis pigmentosa[10]. Furthermore, the presence of cataracts was also identified as a contributing factor to the patient's vision loss, underscoring the complex interplay between these ocular conditions and the overall deterioration of visual function.

In China, the occurrence of glaucoma associated with retinitis pigmentosa (RP) is predominantly characterized by angle-closure types, which are particularly concerning due to their potential for rapid vision loss and the need for timely intervention[11-12]. A comprehensive hospital-centered investigation conducted in China revealed that primary angle-closure glaucoma was present in a striking 30 out of 32 cases examined, constituting a notable 2.3% of the RP patients diagnosed with

glaucoma, which amounted to a total of 32 individuals drawn from a larger cohort of 1,400 RP patients[13]. This statistic underscores the significance of monitoring glaucoma in this specific patient population. Conversely, a contrasting study conducted in Canada indicated that the of primary angle-closure glaucoma among RP patients aged over 40 years was recorded at a much lower rate of 1.03%, highlighting geographical differences in the manifestation of this condition[14]. In the particular case presented, ultrasonographic biomicroscopy (UBM) demonstrated a circumferential narrowing of the angle, which was accompanied by a circumferential angle opening during gonioscopy, while the axial length of the eye remained comfortably within normal suggesting that structural anomalies were not the primary concern [15]. The presence of intraocular hypertension was noted in this patient; consequently, a diagnosis of open-angle glaucoma was established, a condition that is relatively uncommon within the Chinese demographic, further complicating the clinical picture. The precise relationship between RP and glaucoma remains ambiguous and is a subject of ongoing research. Peng's study indicated that there was no pigment deposition observed in the trabecular meshwork (TM), which raises questions about the underlying mechanisms linking these two conditions[13]. Diez-Cattini GF proposed that patients with RP may particularly susceptible to chronic, low-grade inflammatory responses, which could play a role in development of glaucoma[16]. Numerous studies have reported the presence of inflammatory cytokines within the intraocular fluid of patients suffering from both RP and glaucoma, suggesting a potential common pathway that warrants further investigation[17-18]. Additionally, mutations in the IL-20 receptor may influence the capacity of glaucoma patients to adjust outflow resistance in response to elevated intraocular pressure, adding another layer of complexity to the understanding this relationship[19].

We hypothesize that individuals diagnosed with retinitis pigmentosa (RP) may experience inflammation of the trabecular meshwork, which could potentially serve as a contributing factor to the development of open-angle glaucoma, even in cases where there is an absence of pigmentation the aforementioned trabecular meshwork. Furthermore, upon examination, the optic disc appeared be enlarged, a condition that may significantly contribute to the progressive vision loss experienced by these individuals. However, it is important to note that without collaboration in conducting comprehensive visual field assessments, the precise stage of glaucoma in these patients remains undetermined and elusive. The intriguing association between open-angle glaucoma and retinitis pigmentosa warrants additional and thorough investigation to better understand the underlying mechanisms. Future studies could encompass a variety of approaches, including detailed genetic analysis, innovative molecular experimentation, and meticulous pathological evaluations, all aimed at uncovering the complex interplay between these conditions and enhancing our understanding of their relationship.

4. Conclusion

This is a case of peripapillary retinitis pigmentosa with cataract and open-angle glaucoma. The expanded optic cup and cataract may be the reason of vision loss. In RP, open-angle glaucoma may be caused by inflammation reaction.

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Addendum

Abbreviations

RP: retinitis pigmentosa
IOP: intraocular pressure
UBM: Ultrasound biomicroscope
OCT: Optical coherence tomography
RPE: Retinal pigment epithelium
FFA: Fluorescein angiography
OCTA: Optical coherence tomography angiography
ERG: Electroretinography
PSC: posterior subcapsular cataract
TM: trabecular meshwork

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Availability of data and materials

All data generated or analysed during the current study are included in this published article.

Ethics approval and consent to participate

Not applicable. Ethical approval was not required as this manuscript presents a case study.

Consent for publication

The patient gave written permission for clinical details and images in this study. This report does not contain any personal information that could lead to the identification of the patient.

Competing interests

The author declares that they have no competing interests.

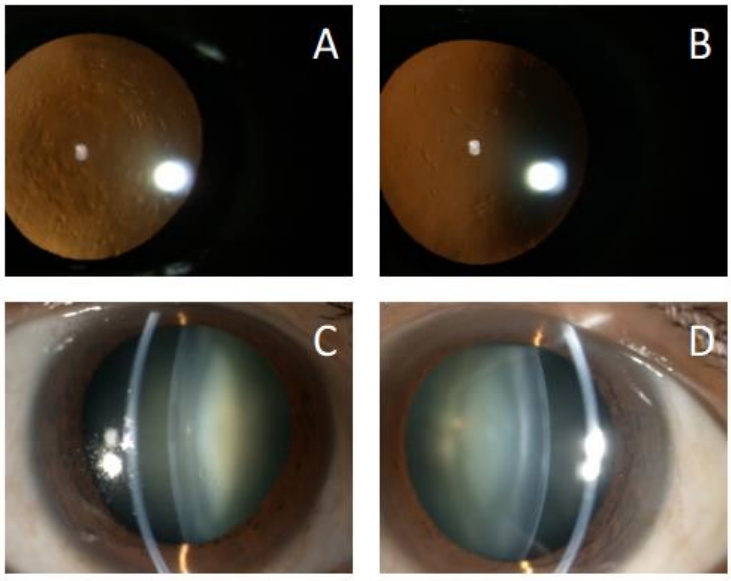

Figure 1: Retroillumination imaging demonstrated posterior subcapsular cataracts (A, B), while slit-lamp examination revealed nuclear opacification (C, D), observed bilaterally.

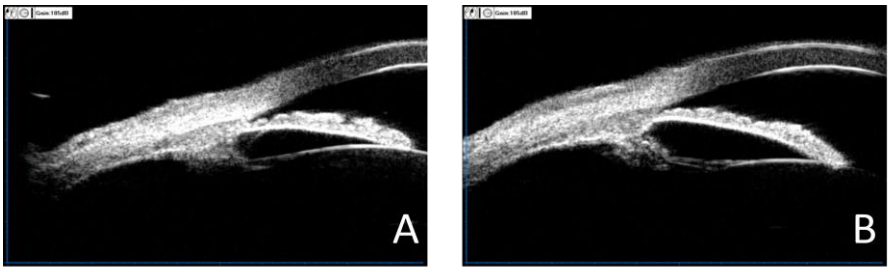

Figure 2: A: UBM findings indicate a narrow angle throughout the entire circumference in the right eye; B: UBM findings indicate a narrow angle throughout the entire circumference in the left eye.

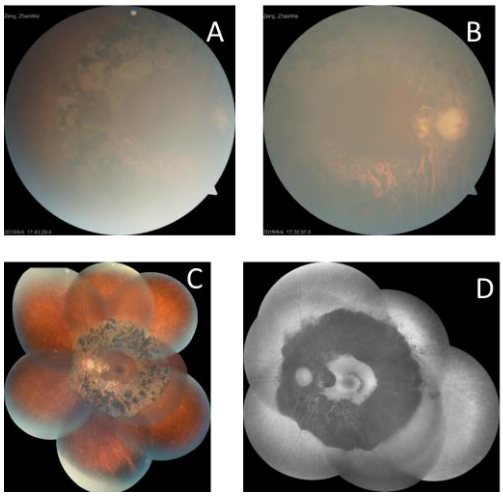

Figure 3: Fundus photography of both eyes showed waxy pallor of the disc, attenuated arterioles, and pigmented atrophic patches perioptic papilla in the right eye(A, B) and in the left eye(C). Autofluorescence highlighted the area of retinal pigment epithelium atrophy (D).

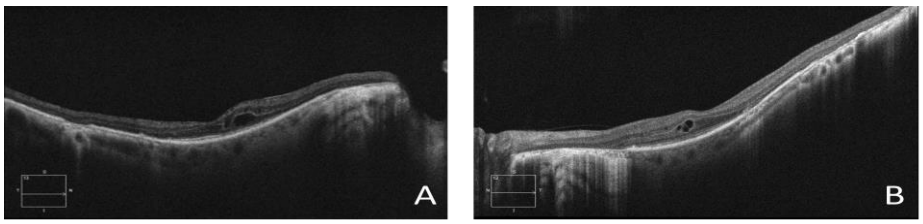

Figure 4: B-Scan optical coherence tomography (OCT) showed thinning of outer retinal layers with the loss of the ellipsoid zone in both eyes (A, B).

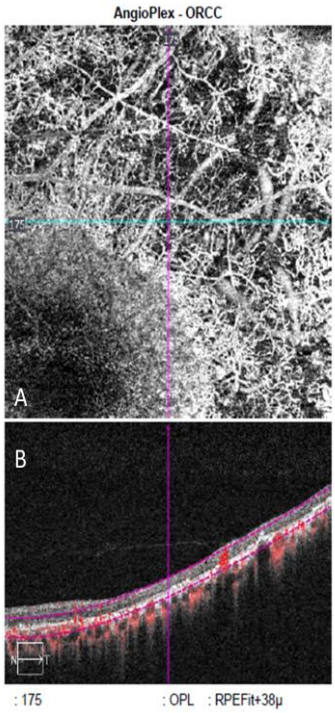

Figure 5: OCTA showed choroid vessels density decreases of the left eye in the region of retinal atrophy.

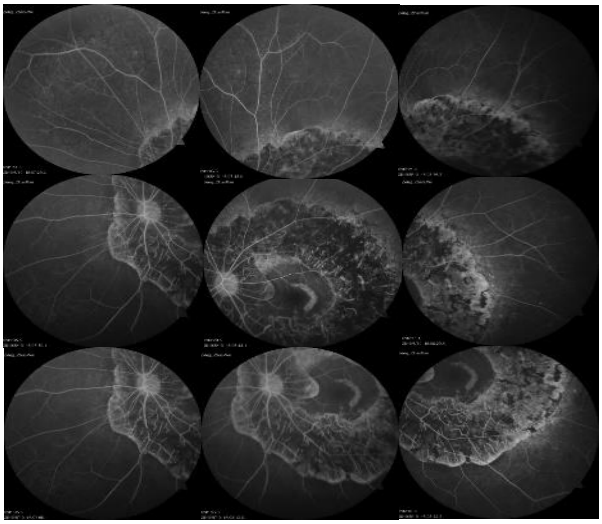

Figure 6: Fluorescein angiography of the left eye showed transmitted fluorescence in early-phase and blocked fluorescence corresponding to the region of atrophy.

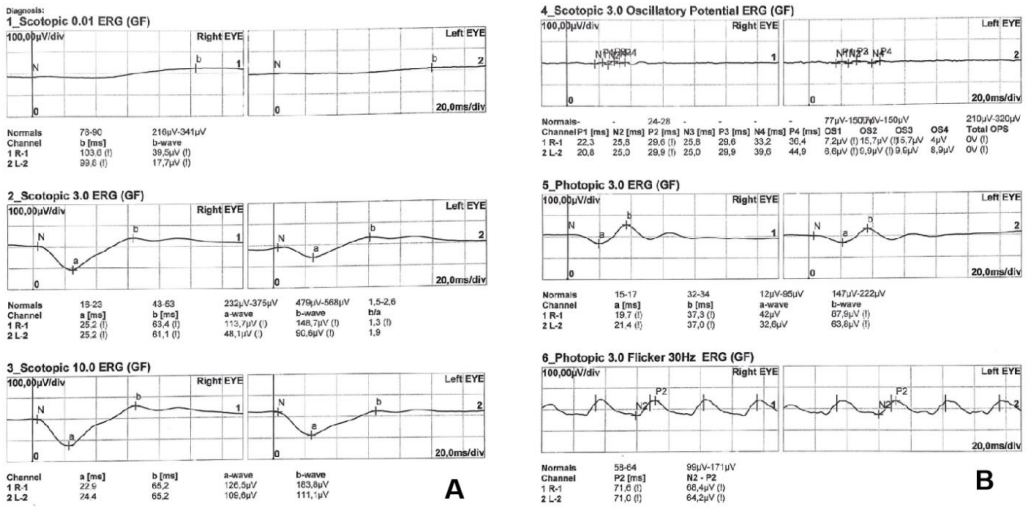

Figure 7: Electroretinography of both eyes showed significant reduction in scotopic and photopic a and b waves and mild reduction in 30Hz flicker response.