

CASE REPORT

Open Access



# Bilateral angle closure glaucoma with retinitis pigmentosa in young patients: case series

Ziyang Lu<sup>1,2,3</sup>, Lu Wang<sup>1</sup>, Xi Ying<sup>1,2\*</sup> and Lian Tan<sup>1,2\*</sup>

## Abstract

**Background** To report the ocular characteristics and management of three cases of retinitis pigmentosa (RP) concurrent primary angle closure glaucoma (PACG).

**Case presentation** Three middle-aged patients presenting with diminished vision, high intraocular pressure (IOP), and typical fundus manifestations of RP were clinically evaluated. The individualized treatment was based on the ocular conditions of each case. A novel genetic alteration in *ZNF408* was identified in one patient. Two patients with short-axial eyes received unilateral combined trabeculectomy, cataract surgery, and Iridio-zonulo-hyaloid-vitreotomy. One of them had a subluxated lens, managed with a capsular tension ring implantation. Their contralateral eyes, respectively, underwent laser peripheral iridotomy (LPI) and transscleral cyclophotocoagulation. The third patient underwent bilaterally combined laser peripheral iridoplasty, LPI, and medication. Ultimately, all patients achieved the target IOP during a two-year follow-up.

**Conclusion** Young patients with RP may have a risk of developing angle closure glaucoma, and conversely, patients with angle closure glaucoma at younger age should be aware of the presence of RP. Therefore, routine gonioscopy and IOP monitoring are required for RP patients, and detailed fundus examinations are warranted for young PACG patients.

**Keywords** Retinitis pigmentosa, Primary angle closure glaucoma, *ZNF408*

## Background

Retinitis pigmentosa (RP), a leading cause of human blindness worldwide, is an inherited retinal degenerated disease [1]. It features progressive loss of photoreceptors and retinal pigment epithelium (RPE). The fundus appearances manifest as waxy optic discs, attenuated retinal vessels, and peripheral pigment migration resembling bony spicules [2]. The full-field electroretinogram (ff-ERG), an essential diagnostic tool for RP, indicates a reduction in a- and b-wave amplitudes. The disease is typically characterized by nyctalopia, decreased visual acuity, and gradual visual field defect, with or without abnormal color vision [1]. The possible explanation of reduced vision in RP can be progressive photoreceptor cell death and treatable complications, including

\*Correspondence:

Xi Ying

yingxieye@163.com

Lian Tan

tanlian0717@126.com

<sup>1</sup> Southwest Hospital / Southwest Eye Hospital, Third Military Medical University (Army Medical University), Chongqing 400038, China

<sup>2</sup> Key Lab of Visual Damage and Regeneration & Restoration of Chongqing, Chongqing 400038, China

<sup>3</sup> Department of Ophthalmology, the 958Th Hospital, Southwest Hospital, Third Military Medical University (Army Medical University), Chongqing 400038, China



© The Author(s) 2023. **Open Access** This article is licensed under a Creative Commons Attribution 4.0 International License, which permits use, sharing, adaptation, distribution and reproduction in any medium or format, as long as you give appropriate credit to the original author(s) and the source, provide a link to the Creative Commons licence, and indicate if changes were made. The images or other third party material in this article are included in the article's Creative Commons licence, unless indicated otherwise in a credit line to the material. If material is not included in the article's Creative Commons licence and your intended use is not permitted by statutory regulation or exceeds the permitted use, you will need to obtain permission directly from the copyright holder. To view a copy of this licence, visit <http://creativecommons.org/licenses/by/4.0/>. The Creative Commons Public Domain Dedication waiver (<http://creativecommons.org/publicdomain/zero/1.0/>) applies to the data made available in this article, unless otherwise stated in a credit line to the data.

cataract, macular abnormalities, glaucoma, and so on. The worldwide prevalence of RP is about 1 in 4000 for a total of more than 1 million patients affected. In previous clinical practices, the co-occurrence of RP and glaucoma has been reported since 1862 [3]. A population-based case–control study in Taiwan showed that RP patients had a 3.64-fold greater odds of developing acute angle closure (ACG) than in the general population [4], supporting a strong association between RP and angle closure glaucoma.

Here, we report three cases of primary angle closure glaucoma (PACG) in RP along with their personalized treatments. Informed consent was obtained from the patients, and all the work conducted was in accordance with the Declaration of Helsinki.

## Case presentations

### Case 1

A 53-year-old female presented with 10 days history of blurry vision in the left eye. She had a family history of RP. On examination, the best-corrected visual acuity (BCVA) was 20/63 OD and only light perception OS. Intraocular pressure (IOP) was 19 mmHg OD and 33 mmHg OS for the first visit, and rising to 27 mmHg OD and 37 mmHg OS one week later, respectively. Pigmentary keratic precipitates (KP), Tyndall phenomenon, dilated pupils with no light response, narrow peripheral anterior chamber (PAC) with Van Herrick (VH) grade 1, and slight lens opacification were observed in bilateral eyes. The presence of osteoblast-like pigmentation in the inferior peripheral retina OU was observed (Fig. S1). The cup to disc ratio (CDR) was nearly 0.5 OD and 0.6 OS (Fig. 1A). No significant abnormalities were evident on the macular optical coherence tomography (OCT) of both eyes (Fig. 1B). The right eye had a visual field defect with retention of a small central and inferotemporal island, while no residual visual field existed in the left eye (Fig. 1C). Gonioscopy of both eyes revealed iridotrabeular synechia in full-circumferential range. A remarkable reduction in a- and b-wave amplitudes was seen in ff-ERG (Fig. S2). We applied genetic testing to the patient and identified a novel single nucleotide homozygous, transversion mutation (c.1618 C > T) of exon 5 in the *ZNF408* gene (Fig. 1E). Both eye received two times daily topical beta-blocker (0.5% timolol), two times daily carbonic anhydrase inhibitors (0.15% brinzolamide), three times daily topical alpha-2 agonists (0.2% brimonidine tartrate) and one times daily travoprost (0.004%). But the left eye still had uncontrolled IOP and consequently undergoing transscleral cyclophotocoagulation (TCP). IOP in the right eye remained well-controlled with topical anti-glaucoma medications for ten months, and became elevated and progressively worsened to a

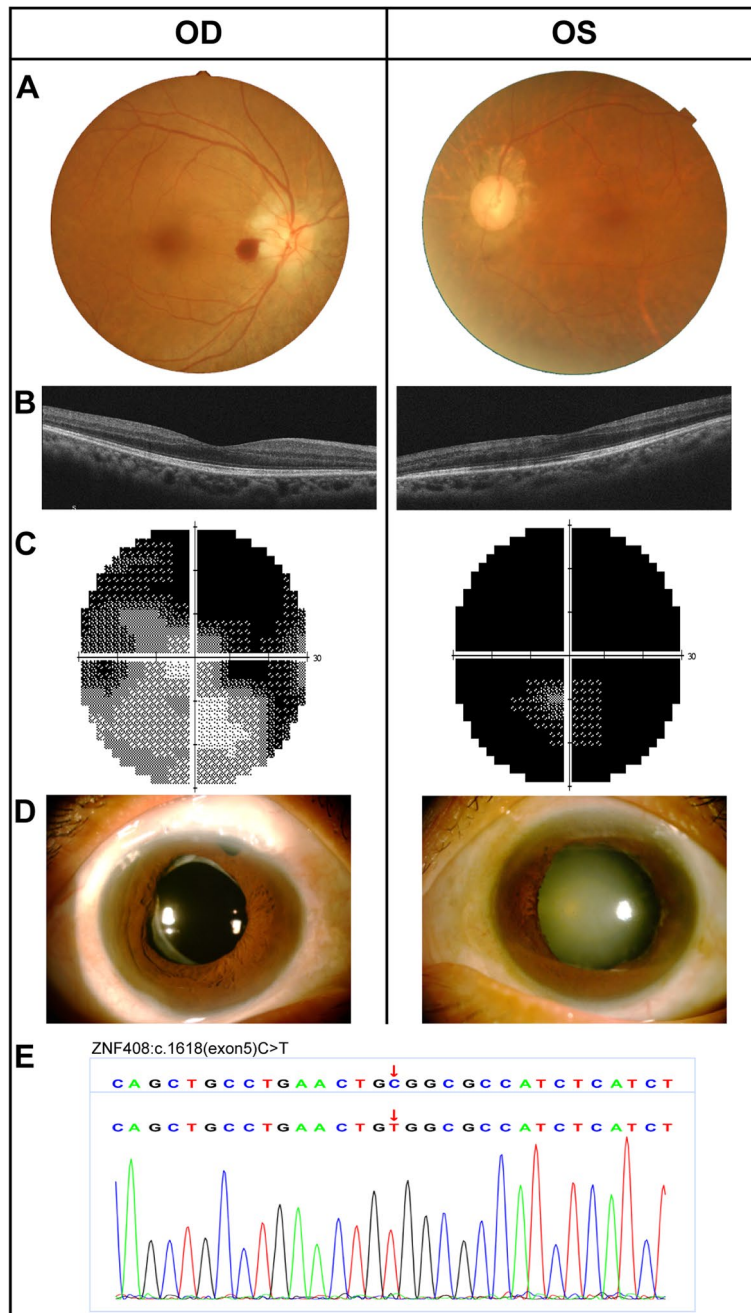
peak of 43 mmHg. Hence, combined cataract surgery, trabeculectomy and the Irido-zonulo-hyaloid-vitreotomy (IZHV) were performed in the right eye. The postoperative anterior segment was kept in a stable condition (Fig. 1D), with BCVA improving to 20/28 OD, and IOP was maintained normal during a 2-year follow-up.

### Case 2

A 54-year-old female presented with a 10 days history of painless visual loss in the right eye. She was otherwise systemically well and had no significant family history. The BCVA was 20/80 OD and 20/40 OS. Goldmann applanation tonometry (GAT) revealed an IOP of 38 mmHg OD and 14 mmHg OS. The depth of PAC was VH grade 1 OU. The pupil was mid-dilated and minimally reacting to light OD. The lens was slightly opacity OU. A fundus examination revealed that discs were waxy pale with a CDR of 0.4 OD and 0.3 OS, respectively. Bony spicule pigmentation diffused in the mid-peripheral and peripheral retina (Fig. 2A). Macular OCT suggested no significant abnormality OU (Fig. 2B). However, a tubular visual field was observed in bilateral eyes (Fig. 2C). Decreased a- and b-wave amplitudes were observed in both eyes with ff-ERG (Fig. S2). Gonioscopy revealed closed angle in all quadrant OD, whereas narrow ACA in three quadrants OS. The ocular biometric parameters of both eyes were shown in Table 1. The IOP was out of control with four kinds of anti-glaucoma medications, thus combined cataract surgery and trabeculectomy were performed on the right eye. Whole-circumference unstable zonular fibers were discovered during phacoemulsification, treated by the implantation of a capsular tension ring. Simultaneously, the right eye underwent the IZHV to prevent malignant glaucoma. Laser peripheral iridotomy (LPI) was performed in the left eye (Fig. 2D). The postoperative BCVA was 20/32 OD, and IOP was maintained within normal at postoperative 2 years.

### Case 3

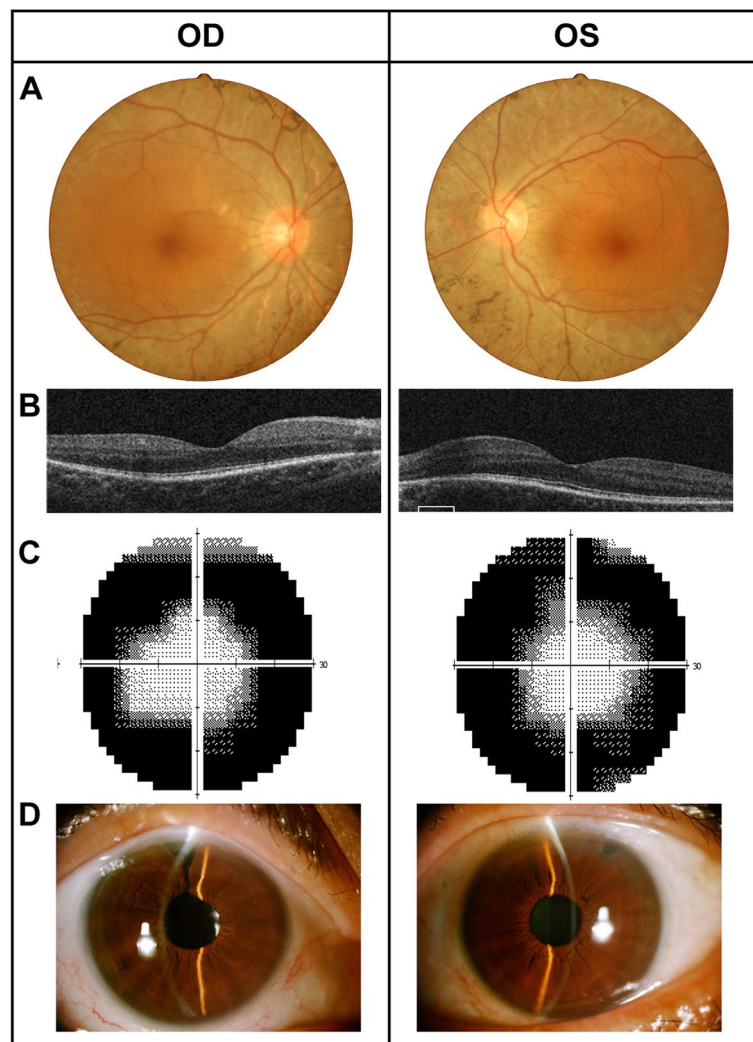
A 47-year-old male with established RP for 13 years was found high IOP bilaterally on the regular reviews. He had a history of high myopia. The BCVA was 20/125 OD and 20/32 OS. IOP with applanation was 27 mmHg OU. A slit-lamp examination showed the peripheral anterior chamber of both eyes was shallow with VH grade 2. The fundus examination revealed the retinal vessels were strongly attenuated, diffused chorioretinal atrophy OU and waxy pale discs with a CDR of 0.9 OD and 0.8 OS. We scarcely found bony spicules existed in the retina (Fig. 3A) and no detectable waves in ff-ERG (Fig. S2). Macular OCT revealed an attenuated outer nuclear layer, RPE, and loss of outer/inner segments (Fig. 3B). Significantly great impairment was observed with



**Fig. 1** Clinical examinations of Case 1. **A** The fundus photography showing a disc hemorrhage OD and a cup to disc ratio of 0.5 OD and 0.6 OS. **B** Macular OCT suggesting no significant abnormality OU. **C** A visual field with retention of a small central and inferotemporal island OD and no residual visual field OS were observed. **D** The anterior segment photograph at 1-year follow-up revealing a functional filtering bleb OD, IOL in place OD, and normal anterior chamber OU. **E** Partial sequence diagram of *ZNF408* exon 5. A homozygous mutation (c.1618 C>T transition)

perimetry (Fig. 3C). Gonioscopy bilaterally indicated the four-quadrant angle was iridotrabecular appositional and could be re-opened in dynamic gonioscope, so combined laser peripheral iridoplasty and LPI were performed in

both eyes. The postoperative PAC widened (Fig. 3D) and IOP consistently was kept under control throughout a 2-year follow-up with the application of topical IOP-lowering eye drops.



**Fig. 2** Clinical examinations of Case 2. **A** Fundus photography of both eye showing waxy pale optic discs and retinal hyperpigmentation in bone spicules at the mid-periphery and periphery retina. **B** Macular OCT showing no significant abnormality OU. **C** Visual field testing demonstrating residual tubular visual field in both eyes. **D** Anterior segment photography at postoperative year 1 revealing a deep anterior chamber of both eyes

### Discussion and conclusions

In this series, we described three uncommon cases of RP concomitant PACG attending our hospital in 2020 and performed a regular follow-up. In our previous work, RP patients from January 2014 to January 2019 were studied retrospectively, which found concurrent glaucoma accounting for 1.6% [5]. Moreover, we reviewed the prevalence of PACG in RP patients, and figured out the Asians appear to rank the highest, especially the Chinese (Table 2). This might be explained by a larger sample size of RP and a higher prevalence in China, approximately 1.6–4.0 times than worldwide [1, 6, 7]. Furthermore, nanophthalmos, cataract, lens subluxation, increased lens thickness (LT) and anterior segment dysgenesis in RP were predisposing factors for ACG anatomically [8, 9]. As

shown in Table 1, two patients had short axial eyes (Axial length < 22 mm). Zonular instability and thicker lens were also observed among them.

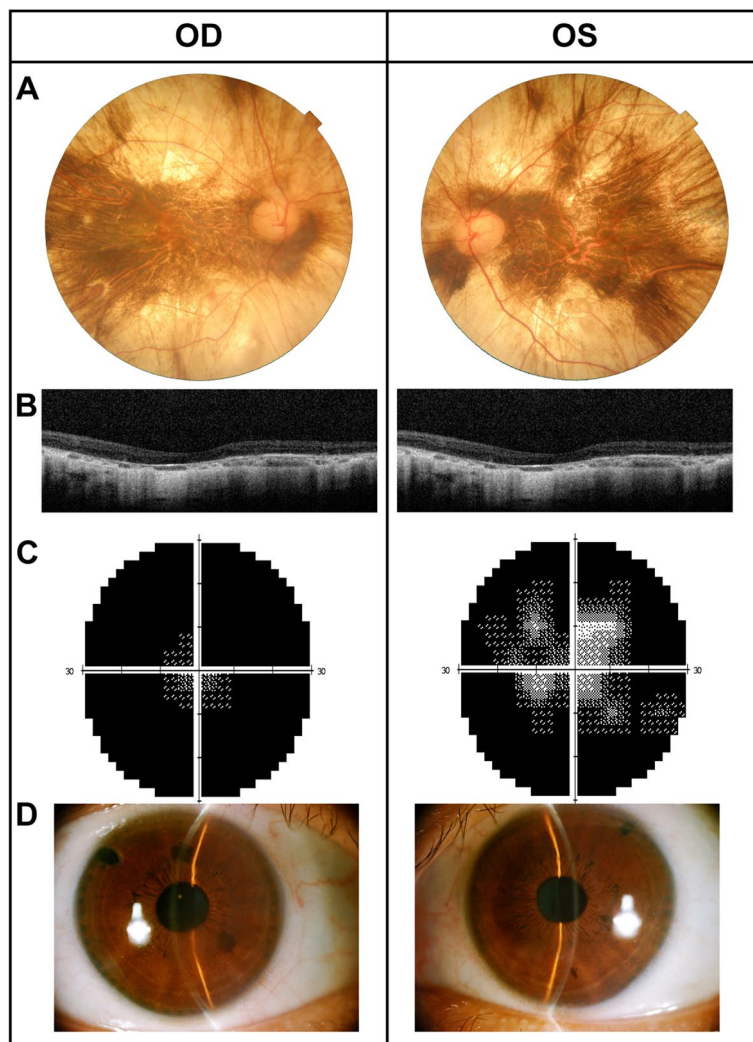
Prevalence of PACG increased with aging steadily. People older than 80 years old have highest prevalence of 2.8%, while people aged 40–49 years have the lowest prevalence of 0.1% [16]. RP patients had an earlier onset of PACG (Table 2) [4, 10–15]. The average age at which PACG is diagnosed in the general population is  $(34.4 \pm 5.1)$  years old, while that in RP patients is  $(29.7 \pm 7.0)$  years old [9]. This was contrary to the fact that PACG was more prevalent in the elderly population [4, 8, 16]. Meanwhile, the incidence of PACG in Chinese RP patients (Table 2) was higher than that in the general middle-aged and elderly (40 years old and

**Table 1** Clinical characteristics and management of the three cases in the study

	Age (yrs)	Gender	AL (mm)	LT (mm)	ACD (mm)	Presenting BCVA	Presenting IOP (mmHg)	Management	2-year postoperative BCVA	2-year postoperative IOP (mmHg)
<b>Case 1</b>	53	Female	OD 21.78 OS 21.80	5.27 5.25	1.73 1.64	20/63 LP	19 33	Trab + Phaco + IOL + AV TCP	20/28 LP	8.6 12.4
<b>Case 2</b>	54	Female	OD 21.75 OS 21.62	4.85 4.64	1.61 2.50	20/80 20/40	38 14	Trab + Phaco + IOL + CTR + AV LPI	20/32 20/40	13 17
<b>Case 3</b>	47	Male	OD 26.63 OS 26.34	5.21 5.06	2.09 2.23	20/125 20/32	27 27	LPIp + LPI LPIp + LPI	20/200 20/40	9.4 9.4

AL Axial length, LT Lens thickness, ACD Anterior chamber depth, BCVA Best Corrected Visual Acuity, LP light perception, Trab trabeculectomy, Phaco phacoemulsification, IOL intraocular lens, TCP transscleral cyclophotocoagulation, CTR capsular tension ring, AV anterior vitrectomy, LPI laser peripheral iridotomy, LPIp laser peripheral iridoplasty





**Fig. 3** Clinical examinations of Case 3. **A** The fundus photography of both eyes with advanced retinitis pigmentosa showing narrowed retinal vessels, extensive chorioretinal atrophy, and waxy pallor of optic discs. **B** Macular OCT of both eyes identifying severe outer retinal layer thinning. **C** Visual field with retention of a small central island was detected in bilateral eyes. **D** Anterior segment photography at postoperative year 1 revealing a wide peripheral anterior chamber bilaterally

**Table 2** Comparison of the prevalence of PACG in RP patients in previous studies

First author (Year of publication)	District	Subjects (No.)	PACG (Subjects /%)	Age <sup>a</sup>
D W Peng (1991) [10]	China	1400	30 (2.14%)	44.0 (range 13–63)
O Badeeb (1993) [11]	Canada	538	5 (0.93%)	57.2 (range 32–74)
Ko YC (2014) [4]	Taiwan, China	382	5 (1.3%)	53.3 ± 8.0
Pradhan C (2020) [12]	Nepal	234	5 (2.13%)	56.6 ± 8.3
Wang DD (2022) [13]	China	1356	39 (2.88%)	46.04 ± 14.50
Hung MC (2022) [14]	Taiwan, China	6223	100 (1.61%)	49.0 ± 18.1
Pradhan ZS (2022) [15]	India	618	14 (2.27%)	52.7 ± 2.4

<sup>a</sup> The average age of the confirmed diagnosis of PACG in RP patients

above) population [16]. Those findings may suggest that a higher risk of developing angle closure glaucoma exist in younger patients with retinitis pigmentosa. However, more than half of the RP patients co-occurred with PACG had a normal optic disc [10]. A tubular visual field might be observed in the patients of RP or PACG at the late stage. It is worth noting that atypical glaucomatous optic discs, similar visual defects, and the incidence of rarity may lead to a higher misdiagnosis rate. Hence, comprehensive ophthalmic examinations, including gonioscopy, ff-ERG, macular OCT and genetic testing, could be warranted in younger RP patients in clinical diagnosis.

Zhong et al.'s exploration using whole exome sequencing discovered that five genes, *CRB1*, *COL2A1*, *RHO*, *RPIL1* and *PAX6*, were responsible for the genetic association between early-onset PACG and RP [17, 18]. Therein, *CRB1* and *RHO* had been implicated in familial forms of nanophthalmos [19–22]. *COL2A1* and *PAX6* were pivotal regulators of anterior segment dysgenesis [23–25]. To our knowledge, here we first report *ZNF408* as a new gene associated with RP concurrent PACG, which was only previously identified alone in familial exudative vitreoretinopathy or RP [26, 27]. *ZNF408* encodes a zinc finger protein that harbors 10 C2H2-type fingers thought to be implicated in DNA binding. *ZNF408* is mostly expressed in human retinas, including photoreceptors, amacrine, ganglion cells and retinal blood vessels [26]. Its mutation could lead to impairment of visual function and typical changes of RP [26]. Remarkably, this is also the first report that the *ZNF408* mutation was identified domestically in RP patients.

The current therapeutic strategies for PACG comprised laser, medication, or surgery (trabeculectomy, lens extraction, combined lens extraction with trabeculectomy, goniosynechialysis, etc.). The management based on cases were listed in Table 1. Notably, two patients underwent anterior vitrectomy. Prior clinical studies demonstrated that the anterior chamber tended to become refractory shallow in PACG patients with retinal disorders after trabeculectomy [28, 29]. Moreover, choroidal expansion secondary to fundus vascular abnormalities, shorter AL and zonular slackness had been proven to increase the incidence of postoperative malignant glaucoma [30, 31]. IZHV has been applied by many ophthalmologists as a golden standard to manage malignant glaucoma [32]. It was also noteworthy that two middle-aged patients without significant lens opacities underwent cataract surgeries. Our previous studies showed that cataract was the second complication in RP patients in Western China, representing approximately 43.1% [5]. Researches concerning RP unambiguously pointed out that inflammation occupied a crucial

position in disease progression [33–35]. Persistent ocular inflammation supposedly damaged lens zonules and induced lens opacity, leading to severe vision impairment and anterior chamber angle occlusion. Therefore, the timing for cataract surgery was recommended to be appropriate earlier for RP patients to alleviate vision impairment and monitor the fundus changes [36–38]. All three patients achieved satisfactory outcomes in a two-year follow-up.

In conclusion, through describing three cases with PR concomitant PACG and their corresponding clinic treatments, we supposed that young patients with RP might have a risk of developing angle closure glaucoma, and conversely, patients with angle closure glaucoma at younger age should be aware of the presence of RP.

#### Abbreviations

RP	Retinitis pigmentosa
PACG	Primary angle-closure glaucoma
IOP	Intraocular pressure
LPI	Laser peripheral iridotomy
RPE	Retinal pigment epithelium
ff-ERG	Full-field electroretinogram
PAC	Peripheral anterior chamber
OCT	Optical coherence tomography
ACA	Anterior chamber angle
TCP	Transscleral cyclophotocoagulation
IZHV	Irido-zonulo-hyaloid-vitrectomy
GAT	Goldmann applanation tonometry
AL	Axial length
LT	Lens thickness
ACD	Anterior chamber depth
BCVA	Best Corrected Visual Acuity
LP	Light perception
Trab	Trabeculectomy
Phaco	Phacoemulsification
IOL	Intraocular lens
CTR	Capsular tension ring
AV	Anterior vitrectomy
LPIp	Laser peripheral iridoplasty

#### Supplementary Information

The online version contains supplementary material available at <https://doi.org/10.1186/s12886-023-03190-y>.

**Additional file 1: Supplementary Figure 1.** The fundus photography of Case 1 showed bony spicule pigmentary changes at the inferior peripheral retina (red rectangle). **Supplementary Figure 2.** The full-field electroretinogram (ff-ERG) of three cases. Scotopic and photopic responses with a slight reduction in the amplitude of a and b waves were observed in Case 1, while nondetectable waves were in Case 2. A remarkable reduction in a- and b-wave amplitudes were seen in Case 3.

#### Acknowledgements

Not applicable.

#### Authors' contributions

ZL: data collection, interpretation and manuscript preparation, critical revision of the article; LW: data collection; LT and XY: contributed equally to the study. Investigation design, manuscript preparation and critical revision of manuscript. All authors have read and approved the manuscript and are equally accountable for all aspect of this work.

**Funding**

This work was supported by the Chongqing Science and Technology Commission of China (cstc2020jcyj-msxmX1097).

**Availability of data and materials**

Data could be available from the corresponding author by reasonable inquire.

**Declarations****Ethics approval and consent to participate**

This study was performed according to the Declaration of Helsinki and approved by the Ethics and Research Committee of Southwest Hospital, Army Medical University. Written consent from the patients was not necessary for this retrospective descriptive study of case series.

**Consent for publication**

Informed consent was obtained from all the patients for publication of this case report and any accompanying images.

**Competing interests**

The authors declare no competing interests.

Received: 28 May 2023 Accepted: 27 October 2023

Published online: 15 November 2023

**References**

- Hartong DT, Berson EL, Dryja TP. Retinitis pigmentosa. *Lancet*. 2006;368(9549):1795–809.
- Dryja TP, McGee TL, Reichel E, Hahn LB, Cowley GS, Yandell DW, et al. A point mutation of the rhodopsin gene in one form of retinitis pigmentosa. *Nature*. 1990;343(6256):364–6.
- Wang M, Lin HT, Bai YJ, Ge J, Zhuo YH. Clinical evidence in concurrence of retinitis pigmentosa and glaucoma. *Chin Med J (Engl)*. 2011;124(8):1270–4.
- Ko YC, Liu CJ, Hwang DK, Chen TJ, Liu CJ. Increased risk of acute angle closure in retinitis pigmentosa: a population-based case-control study. *PLoS One*. 2014;9(9):e107660.
- Tan L, Long Y, Li Z, Ying X, Ren J, Sun C, et al. Ocular abnormalities in a large patient cohort with retinitis pigmentosa in Western China. *BMC Ophthalmol*. 2021;21(1):43.
- Xu L, Hu L, Ma K, Li J, Jonas JB. Prevalence of retinitis pigmentosa in urban and rural adult Chinese: the Beijing Eye Study. *Eur J Ophthalmol*. 2006;16(6):865–6.
- Teo CL, Cheung N, Poh S, Thakur S, Rim TH, Cheng CY, et al. Prevalence of retinitis pigmentosa in Singapore: the Singapore epidemiology of eye diseases study. *Acta Ophthalmol*. 2021;99(1):e134–5.
- Xu J, Ouyang Z, Yang Y, Cai X, Wang Z, Lin M, et al. Ocular biometry in primary angle-closure glaucoma associated with retinitis pigmentosa. *J Ophthalmol*. 2017;2017:9164846.
- Gao F, Wang J, Chen J, Wang X, Chen Y, Sun X. Etiologies and clinical characteristics of young patients with angle-closure glaucoma: a 15-year single-center retrospective study. *Graefes Arch Clin Exp Ophthalmol*. 2021;259(8):2379–87.
- Peng DW. Retinitis pigmentosa associated with glaucoma. [Zhonghua yan ke za zhi] *Chin J Ophthalmol*. 1991;27(5):262–4.
- Badeeb O, Trope G, Musarella M. Primary angle closure glaucoma and retinitis pigmentosa. *Acta Ophthalmol*. 1993;71(6):727–32.
- Pradhan C, Khadka S, Joshi P. Angle closure glaucoma in retinitis pigmentosa. *Case Rep Ophthalmol Med*. 2020;2020:6023586.
- Wang DD, Gao FJ, Hu FY, Cao WJ, Xu P, Huang Y, et al. Clinical and genetic analysis of retinitis pigmentosa with primary angle closure glaucoma in the Chinese population. *Curr Eye Res*. 2022;47(9):1339–45.
- Hung MC, Chen YY. Association between retinitis pigmentosa and an increased risk of primary angle closure glaucoma: a population-based cohort study. *PLoS One*. 2022;17(9):e0274066.
- Pradhan ZS, Shroff S, Bansod A, Poornachandra B, Shetty A, Devi S, et al. Prevalence of primary angle-closure disease in retinitis pigmentosa. *Indian J Ophthalmol*. 2022;70(7):2449–51.
- Zhang N, Wang J, Chen B, Li Y, Jiang B. Prevalence of primary angle closure glaucoma in the last 20 years: a meta-analysis and systematic review. *front Med*. 2020;7:624179.
- Liu X, Li J, Lin S, Xiao X, Luo J, Wei W, et al. Evaluation of the genetic association between early-onset primary angle-closure glaucoma and retinitis pigmentosa. *Exp Eye Res*. 2020;197:108118.
- Xiao T, Xu K, Zhang X, Xie Y, Li Y. Sector Retinitis Pigmentosa caused by mutations of the RHO gene. *Eye*. 2019;33(4):592–9.
- Zenteno JC, Buentello-Volante B, Ayala-Ramirez R, Villanueva-Mendoza C. Homozygosity mapping identifies the Crumbs homologue 1 (Crb1) gene as responsible for a recessive syndrome of retinitis pigmentosa and nanophthalmos. *Am J Med Genet A*. 2011;155A(5):1001–6.
- Paun CC, Pijl BJ, Siemiatkowska AM, Collin RW, Cremers FP, Hoyng CB, et al. A novel crumbs homolog 1 mutation in a family with retinitis pigmentosa, nanophthalmos, and optic disc drusen. *Mol Vis*. 2012;18:2447–53.
- Ehrenberg M, Pierce EA, Cox GF, Fulton AB. CRB1: one gene, many phenotypes. *Semin Ophthalmol*. 2013;28(5–6):397–405.
- Yu Y, Xia X, Li H, Zhang Y, Zhou X, Jiang H. A new rhodopsin R135W mutation induces endoplasmic reticulum stress and apoptosis in retinal pigment epithelial cells. *J Cell Physiol*. 2019;234(8):14100–8.
- Halder G, Callaerts P, Gehring WJ. Induction of ectopic eyes by targeted expression of the eyeless gene in *Drosophila*. *Science*. 1995;267(5205):1788–92.
- Shaham O, Menuchin Y, Farhy C, Ashery-Padan R. Pax6: a multi-level regulator of ocular development. *Prog Retin Eye Res*. 2012;31(5):351–76.
- Richards AJ, Martin S, Yates JR, Scott JD, Baguley DM, Pope FM, et al. COL2A1 exon 2 mutations: relevance to the Stickler and Wagner syndromes. *Br J Ophthalmol*. 2000;84(4):364–71.
- Avila-Fernandez A, Perez-Carro R, Corton M, Lopez-Molina MI, Campello L, Garanto A, et al. Whole-exome sequencing reveals ZNF408 as a new gene associated with autosomal recessive retinitis pigmentosa with vitreal alterations. *Hum Mol Genet*. 2015;24(14):4037–48.
- Collin RW, Nikopoulos K, Dona M, Gilissen C, Hoischen A, Boonstra FN, et al. ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. *Proc Natl Acad Sci USA*. 2013;110(24):9856–61.
- Zhong Y, Guo X, Xiao H, Luo J, Zuo C, Huang X, et al. Flat anterior chamber after trabeculectomy in secondary angle-closure glaucoma with BEST1 gene mutation: case series. *PLoS One*. 2017;12(1):e0169395.
- Li M, Zhu W, Fan X, Sun X, Kong X. Outcomes of filtering surgery versus clear lens extraction in young patients with angle-closure glaucoma. *Am J Ophthalmol*. 2023;S0002-9394(23):00314–8.
- Quigley HA, Friedman DS, Congdon NG. Possible mechanisms of primary angle-closure and malignant glaucoma. *J Glaucoma*. 2003;12(2):167–80.
- Chandler PA, Grant WM. Mydriatic-cycloplegic treatment in malignant glaucoma. *Arch Ophthalmol*. 1962;68:353–9.
- Yu X, Zhao Z, Zhang D, Yang X, Sun N, Lin Y, et al. Anterior vitrectomy, phacoemulsification cataract extraction and irido-zonulo-hyaloid-vitrectomy in protracted acute angle closure crisis. *Int Ophthalmol*. 2021;41(9):3087–97.
- Singh I, Singh AK, Contreras MA. Peroxisomal dysfunction in inflammatory childhood white matter disorders: an unexpected contributor to neuropathology. *J Child Neurol*. 2009;24(9):1147–57.
- Vitner EB, Platt FM, Futerman AH. Common and uncommon pathogenic cascades in lysosomal storage diseases. *J Biol Chem*. 2010;285(27):20423–7.
- McMurtrey JJ, Tso MOM. A review of the immunologic findings observed in retinitis pigmentosa. *Surv Ophthalmol*. 2018;63(6):769–81.
- Jackson H, Garway-Heath D, Rosen P, Bird AC, Tuft SJ. Outcome of cataract surgery in patients with retinitis pigmentosa. *Br J Ophthalmol*. 2001;85(8):936–8.
- Yoshida N, Ikeda Y, Murakami Y, Nakatake S, Fujiwara K, Notomi S, et al. Factors affecting visual acuity after cataract surgery in patients with retinitis pigmentosa. *Ophthalmology*. 2015;122(5):903–8.
- Bayyoud T, Bartz-Schmidt KU, Yoeruek E. Long-term clinical results after cataract surgery with and without capsular tension ring in patients with retinitis pigmentosa: a retrospective study. *BMJ open*. 2013;3(4):e002616.

**Publisher's Note**

Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.