Novel genetic mutations in genes AGBL5 and TULP1 for presumed unilateral retinitis pigmentosa managed with low vision rehabilitation: A case report and review

Maggie Man Ki Ho OD, MS, FAAO
University of Incarnate Word, maggs_nunavut@hotmail.com

Stephanie Schmiedecke-Barbieri OD, FAAO, ABCMO, Dip Low Vision
University of Incarnate Word, schmiede@uiwtx.edu

Patricia C. Sanchez-Diaz PhD, DVM, FAAO
University of the Incarnate Word, sanchezd@uiwtx.edu

Carolyn E. Majcher OD, FAAO, FORS
Northeastern State University, majcher@nsuok.edu

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Novel genetic mutations in genes AGBL5 and TULP1 for presumed unilateral retinitis pigmentosa managed with low vision rehabilitation: A case report and review

Abstract

Background: Retinitis pigmentosa is a group of hereditary retinal diseases characterized by the degeneration of rod and cone photoreceptors. It commonly results in night blindness followed by tunnel vision and central vision reduction. The classic triad of clinical signs includes pigmented bone spicules, waxy disc pallor, and arterial attenuation. Unilateral retinitis pigmentosa is rare and can be supported with ancillary testing including genetic and laboratory studies to rule out differential diagnoses.

Case Report: A 68-year-old Hispanic female was referred to the low vision rehabilitation clinic due to progressive vision loss in the left eye (OS) that began 15 years ago. The vision was normal in the right eye (OD). Additionally, she suffered from hearing loss in the right ear since age 3. Examination revealed abnormal visual acuity, visual field, fundus appearance, optical coherence tomography, and electrodiagnostic test results in the OS only. Laboratory studies ruled out various infectious, autoimmune, traumatic, and toxic drug etiologies. Genetic testing revealed novel mutations in genes associated with retinitis pigmentosa.

Conclusion: The genetic testing results along with the clinical examination and electrodiagnostic evaluation supports the diagnosis of unilateral retinitis pigmentosa.

Keywords
unilateral retinitis pigmentosa, genetic, low vision, AGBL5, TULP1

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Cover Page Footnote

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INTRODUCTION

Retinitis pigmentosa (RP) is a genetically inherited group of disorders affecting 1 in 5,000 people worldwide and 1 in 4,000 in the United States.\(^1\) It is commonly bilateral with a variety of presentations and progressions. The initial symptom is night blindness followed by reduced peripheral visual field. During the later stages of the disease, color and central vision become affected.\(^2\) Characteristic fundus features include pigmented bone spicules, waxy disc pallor, and arterial attenuation. Other ocular findings include cystoid macular edema and posterior subcapsular cataract. The genetic inheritance pattern of RP varies, and may be autosomal dominant, autosomal recessive, X-linked recessive, or sporadic.\(^1\)

Although there is no cure for retinitis pigmentosa, there are options for supportive care. One example is nutriceutical vitamin supplementation. Research has shown that taking a high dose of vitamin A palmitate (15,000 IU daily), omega-3 rich foods, and a lutein supplement may slow the progression of RP.\(^2,3\) Additional prophylactic measures include protective sunglasses and avoiding smoking, since exposure to ultraviolet light and smoking increases oxidative damage and accelerates retinal degeneration.\(^3\) If sequelae occur, such as cystoid macular edema, off-label carbonic anhydrase inhibitors may be considered and have been reported to have a response rate of 40 to 50%.\(^4,5\)

Gene and stem cell therapies are two exciting and rapidly evolving areas of research. For those patients with \textit{RPE65} mutations, a novel gene therapy, Luxturna®, was approved in December of 2017 by the Food and Drug Administration (FDA). This was the first FDA approved gene therapy for an inherited disease in the United States.\(^2,6,7\) Gene and stem cell therapies may slow retinal degeneration in RP and may also improve visual function.\(^8\) Several clinical trials targeting other genetic mutations for RP are currently underway.\(^8,9\)

RP is a heterogenous group of retinal dystrophies with several sequelae resulting from different subtypes described. Reclassification of some of these variants is likely as we uncover the molecular pathogenesis and define different presentations of the disease. Some clinical differentials of RP include:

- \textit{Minor asymmetrically bilateral RP:} The common form of RP is bilateral, but the disease progresses faster in one eye compared to the fellow eye. This results in minor asymmetry in RP presentation.\(^10\)
- \textit{Unilateral RP (URP):} The first case of URP was described in 1948 by Dreisler.\(^10\) Since then, nearly 100 cases of URP have been reported based on the following criteria established by Francois and Verriest: (i) unilateral ocular RP
presentation, (ii) normal fundus appearance and electroretinogram (ERG) findings in the fellow eye, (iii) exclusion of infectious etiology, and (iv) sufficiently long period of observation (more than 5 years). These criteria have been supported by later studies, however, there is still controversy concerning the nature of this clinical presentation.

- **Pseudo-RP**: There are several etiologies that can mimic RP including infection (i.e., syphilis, toxoplasmosis, rubella, chicken pox, measles, cytomegalovirus), inflammation (i.e., uveitis, retinal vasculitis), autoimmunity (i.e., cancer-associated retinopathy), trauma (i.e., blunt trauma, retinal detachment), or drug toxicity (i.e. chloroquine/hydroxychloroquine, phenothiazines, thioridazine).

From a low vision standpoint, there are options available throughout the course of vision loss when the condition presents bilaterally. For patients with early vision loss, non-optical aids can be helpful. Some of these include a flashlight for night mobility and a mobility cane. Optically, reverse telescopes to enhance the peripheral field can be helpful in addition to magnification devices for central vision loss. In the past, for patients with end-stage RP (i.e., light perception or worse), an Argus II Retinal Prosthesis implant was considered. This implant sent a signal downstream of the affected retina in the visual pathway, restoring some movement detection and resolution of shapes. (This device is limitedly available.) Regardless of the stage of vision loss and treatment, patients with RP will benefit from management through a coordinated team effort.

In this manuscript we describe the clinical and molecular findings in a patient with a unilateral and progressive form of retinal degeneration. We will also discuss a potential diagnosis of unilateral RP based on clinical history and genetic findings. Although our patient did not exhibit bilateral loss, a brief explanation of how low vision rehabilitation can help in cases of bilateral retinitis pigmentosa will also be presented.

**CASE REPORT**

A 68-year-old Hispanic female was referred to the low vision rehabilitation clinic due to progressive vision loss of her left eye (OS) that began 15 years prior. She had no visual complaints regarding her right eye (OD). She reported that her OS had fundus findings compatible with RP in her early 40’s.

Her medical history included hypercholesterolemia, type 2 diabetes mellitus diagnosed 6 months ago (uncertain if controlled at initial presentation), and hearing loss in the right ear diagnosed when she was 3 years old (presumably associated
with maternal rubella). She denied any history of trauma, retinal detachment, infectious or inflammatory disease. She had no known allergies. Medications included ibuprofen 600 mg, aspirin 81 mg, atorvastatin 20 mg, lisinopril 5 mg, omeprazole 20 mg, and captopril 25 mg. She was not on diabetic medications. She was a former occasional smoker but denied alcohol consumption or recreational drug use. Her family history included a paternal uncle with vision loss of unknown etiology.

At presentation, her best-corrected visual acuities were 20/20 OD and 20/40 OS. Contrast sensitivity testing showed slight reduction in OD (log 1.50) and severe reduction OS (log 0.75). Pupil testing revealed a 3 to 4+ relative afferent pupillary defect OS. A 120-point screening automated Humphrey visual field demonstrated 115/120 points seen OD and 0/120 points seen OS with a size III white stimulus (Figure A.1a).

![Figure A.1a. 120 point visual field showing an essentially full field OD, and near-total visual field loss OS. The HVF 30-2 shows some central vision sparring OS.](image)

Fundus evaluation revealed senescent macular changes and atypical scattered granular white dots throughout the midperiphery in both eyes (Figures A.2, OD and A.3 for OS). In the OD, extensive drusenoid deposits were present in the macula and posterior pole consistent with intermediate stage nonexudative age-related macular degeneration (Figure A.2).
Figure A.2. Fundus photo OD showing drusen throughout the macula and posterior pole.

Figure A.3. Two fundus photos of OS showing patchy areas of RPE atrophy, bone-spicule shaped RPE hyperplasia, arteriolar attenuation, choroidal vessel sclerosis, optic atrophy, and mild macular drusen.

The optic nerve head appeared normal with a 0.3 cup-to-disc ratio and the retinal vasculature was unremarkable aside from a mildly enlarged arteriolar light reflex. Minor pigmentary changes were present OD, and only a single locus of retinal pigment epithelium (RPE) hyperplasia was present along a retinal vessel in the nasal midperiphery. Fundus autofluorescence (FAF) imaging OD revealed pinpoint scattered speckled or granular mixed hypofluorescent and hyperfluorescent changes throughout the posterior pole (Figure A.4).
Few large-sized drusen in the macular region corresponded to rings of hyperfluorescence with central hypofluorescence. Additionally, a ring of hypofluorescence surrounding the optic nerve head was present consistent with nonpathologic peripapillary atrophy. Structural optical coherence tomography (OCT) OD confirmed the presence of large-sized soft drusenoid deposits within the macular region. OCT angiography OD demonstrated normal appearing inner retinal vasculature, however, mild focal perfusion deficits were present within the choriocapillaris underlying areas of drusen (Figure A.5). No signs of exudative age-related macular degeneration (such as fluid or choroidal neovascularization) were present with OCT and OCT angiography imaging.
Ophthalmoscopy OS revealed mild drusenoid deposits within the macula and scattered throughout the posterior pole, however, drusenoid changes were far less extensive OS compared to OD (Figure A.3). Patchy areas of RPE atrophy and classic appearing bone-spicule shaped RPE hyperplasia were present throughout the posterior pole and periphery OS. The retinal vasculature demonstrated severe diffuse arteriolar attenuation and moderate attenuation of the retinal veins. The large choroidal vessels had a whitened appearance consistent with vascular sclerosis. Severe and diffuse pallor of the neuroretinal rim tissue without cupping was present OS. The cup-to-disc ratio was measured as 0.4. FAF imaging OS revealed patchy and mostly geographic-shaped areas of hypofluorescence that were confluent surrounding the optic nerve (Figure A.4). Additionally, mild relative hyperfluorescence appeared to be present in the remaining macular and foveal regions. OCT angiography OS revealed decreased perfusion in both the retinal and choriocapillaris circulatory systems (Figure A.5). Near total choriocapillaris dropout was present underlying circular geographic areas of RPE loss within the superior macula (Figure A.5). Structural OCT imaging OS demonstrated outer retinal and RPE atrophy with loss of the ellipsoid zone, or photoreceptor integrity line, throughout the macula that spared the foveal region. Few hyperreflective hard drusenoid-type subretinal deposits were present (Figure A.6). Optic nerve head and nerve fiber layer (NFL) OCT imaging showed probable inferior and superior NFL loss OS as well as a relative decrease in NFL thickness OS as compared to OD (Figure A.7).

Figure A.6. Structural macular OCT imaging OS demonstrating outer retinal and RPE atrophy with loss of the ellipsoid zone throughout the macula that spares the center of the fovea.
A series of electrodiagnostic tests were performed to evaluate retinal function. A Diopsys® full-field ERG was obtained utilizing a red flash on a blue background. This waveform appeared normal OD, however, the response OS was nearly extinguished with reduced A-wave, B-wave and photopic negative response amplitudes (Figure A.8).
Figure A.8. Diopsys® full-field ERG obtained utilizing a red flash on a blue background showing normal response OD and near extinguished response OS.

Similarly, Diagnosys® dark-adapted full-field flash ERGs revealed normal A and B-waves OD, and a severely reduced response OS (Figure A.9). Focusing on more localized areas of reduction, the multifocal ERG demonstrated mild reduction in the OD and severe reduction in the OS responses. A contrast sensitivity ERG (which detects functional performance of retinal ganglion cells) showed normal response OD and reduced response OS.
Figure A.9. Diagnosys® light-adapted flicker ERG (left) and dark-adapted full-field flash ERG (right). Dark-adapted full-field flash ERG shows normal A and B-waves OD, and near extinguished response OS.

Additional information including bloodwork and carotid ultrasound imaging was ordered to rule out any attributable infectious, inflammatory, autoimmune, or toxic retinal disease process. The following test results were within normal limits (summarized on Table B.1): complete blood count (CBC), comprehensive metabolic panel, erythrocyte sedimentation rate (ESR), rapid plasma reagin (RPR), fluorescent treponemal antibody absorption (FTA-ABS), Lyme antibody, toxoplasma antibody, and lupus panel. However, C-reactive protein (CRP) was elevated at 20.0 mg/L, outside the normal range of < 8 mg/L, and glucose and blood urea nitrogen (BUN) to creatinine ratio were also flagged as high. Blood glucose was 116 mg/dL, outside the normal range of 65-99 mg/dL, consistent with her history of type 2 diabetes. Because she had hypercholesteremia, we attributed the elevated CRP to be an inflammatory marker for associated cardiovascular disease. Her most current carotid ultrasound revealed patent carotid and vertebral arteries bilaterally.

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Table B.1. Laboratory studies showing within normal levels (WNL) values for all components except C-Reactive Protein (CRP), glucose, and BUN/creatin ratio.
To further assist in the diagnosis, the patient underwent genetic testing via a research program sponsored by the Foundation Fighting Blindness. Sequence analysis using the Blueprint Genetics (BpG) Retinal Dystrophy Panel Plus identified heterozygous missense mutations in AGBL5 [c.14466G>A, p. (Arg489His)] and TULP1 [c.38C>A, p. (Ala13Asp)] genes. These genetic changes were classified as Variants of Uncertain Significance (VUS) because they are inherited with an autosomal recessive pattern and thus, in heterozygosity, were not consistent with a diagnosis of bilateral RP. The patient was educated on RP and counseled on the genetic findings.

While URP did not necessitate low vision rehabilitation, the co-morbidity of AMD with advanced peripheral field loss OS did. So, management with low vision rehabilitation was implemented. Recommendations included a yellow filter to subjectively enhance contrast sensitivity binocularly as both eyes had minimal reduction in contrast sensitivity, and good, direct illumination on her tasks to enhance contrast. To address the near total field loss on her left side, we provided training on scanning her environment to the left and wearing single vision distance spectacles. It was recommended that she perform a behind the wheel driving evaluation with the Department of Motor Vehicles as well. This was suggested as a protective measure for the patient as the state driving law has a recommended, thought not required, minimum horizontal angle of vision.

**DISCUSSION**

URP is a rare disease that usually presents more commonly in adults.\textsuperscript{14,15} It has later onset than bilateral RP and a definitive diagnosis requires thorough investigation including confirmation of normal ERG tests and exclusion of asymmetrical RP as well as other infectious, inflammatory, traumatic, toxic, and vascular retinal conditions.\textsuperscript{14,19} For this patient the clinical presentation, progression of vision loss, and the results of ancillary tests were consistent with URP and fit most of the criteria for URP diagnosis proposed by Francois and Verriest.\textsuperscript{11,15} These criteria include: (i) unilateral ocular RP presentation, (ii) normal fundus appearance and electroretinogram (ERG) findings in the fellow eye, (iii) exclusion of infectious etiology, and (iv) sufficiently long period of observation (more than 5 years). In our patient, fundus findings consistent with RP were present in the OS, with only a single locus of RPE hyperplasia present along a retinal vessel in the nasal midperiphery in the OD. ERG testing was normal OD and laboratory testing ruled out an attributable infectious disease. Additionally, our patient was diagnosed with RP in the OS approximately 25 years prior to her examinations at our clinic.
There are characteristic fundus features and ancillary test findings that aid with the diagnosis of RP. In our patient, fundoscopy revealed the pigmentary bone spicules, arteriolar attenuation, and disc pallor in the OS which directed us to a diagnosis of RP. The bilateral drusen was worse OD, and is indicative of coexisting nonexudative age-related macular degeneration. Otherwise, the fundus appearance OD lacked the distinctive signs of RP. ERG provided a quantitative objective measurement of retinal function\(^2,20\) with photopic and scotopic ERG results severely reduced in the affected eye and normal in the fellow eye, as expected in an individual with URP.

Fundus autofluorescence imaging reflects retinal metabolism as well as the amount and distribution of lipofuscin.\(^21\) Therefore, hypofluorescence is suggestive of disruption or loss of the photoreceptors and/or RPE, while hyperfluorescence is suggestive of retinal stress and impending degeneration. It often reveals even subtle RPE damage that may be difficult to detect with funduscopic examination alone. Fundus autofluorescence imaging in our patient helped highlight widespread and large areas of RPE atrophy that spared the foveal region OS and supported our presumed diagnosis of RP. Her OS also exhibited mild hyperfluorescence within the remaining macular/foveal region which is a well-described FAF feature in eyes with RP.\(^22\) In contrast, FAF imaging OD lacked characteristic features of RP such as midperipheral hypofluorescence and macular hyperfluorescence. The diffuse stippled hyper/hypofluorescence pattern present in the posterior pole OD is likely attributable to nonexudative age-related macular degeneration.

Optical coherence tomography demonstrated outer retinal and RPE atrophy with relative foveal sparing OS only, which is consistent with URP. The intact foveal ellipsoid zone OS is consistent with the relatively good visual acuity. Asymmetric decreased choriocapillaris perfusion, more severe in the OS, was visualized by OCT angiography imaging and is likely attributable to age-related macular degeneration in her OD alone and the combination of advanced RP and age-related macular degeneration in her OS. Additionally, the retinal perfusion was also drastically reduced OS (compared to OD) which is consistent with URP, since research suggests that both reduced retinal and choriocapillaris perfusion are manifestations of RP.\(^23\)

The patient experienced moderate hearing impairment in one ear with an intact vestibular system diagnosed early in life. Her symptoms did not correlate with Usher syndrome or other types of syndromic RP. (Usher syndrome is classified into three subtypes including USH type I, type II, and type III.\(^24,25\) Type I is the most severe, presenting with profound to total deafness, vestibular dysfunction, and progressive RP at birth. Type II is as common, but not as severe as type I, presenting
with variable degree of hearing impairment, intact vestibular system, and RP onset at puberty. Type III presents with moderate deafness, inconsistent vestibular dysfunction, and RP symptoms.) The patient’s mother was infected with rubella during pregnancy, and there is ample evidence indicating that maternal rubella can lead to hearing loss and retinopathy either bilaterally or unilaterally. However, the patient had congenital hearing loss of the right ear while retinopathy was diagnosed only during her fifth decade of life in the contralateral eye. Given this, maternal rubella could explain her congenital hearing loss but it is unlikely related to her retinal disease.

Pseudo-RP due to infection, autoimmunity, trauma, or drug toxicity were ruled out with laboratory testing. The patient showed augmented CRP levels, which may be related to her hypercholesterolemia or uncontrolled diabetic condition (glucose level of 116 mg/dL; normal range of 65-99 mg/dL). However, an inflammatory pseudo-RP could not be completely ruled out in this patient and would require further investigation.

The association between AGBL5 and retinitis pigmentosa 75 was proposed in 2015 and new genetic variants have been discovered since then.27,28 AGBL5 encodes ATP/GTP binding protein like 5 involved in the process of deglutamylation.29 TULP1 gene is involved in photoreceptor function and the lifespan of photoreceptor cells.30 Bilateral RP can be caused by homozygous or compound heterozygous mutations in the autosomal genes AGBL5 and TULP1.27,29,31 In this case, genetic testing found missense mutations in only one of the two alleles of these RP-associated genes (heterozygous expression). Whether or not these changes were causing RP in this case remains elusive. Three possibilities could explain the pathogenic expression of these gene variants: (I) low level mosaicism, defined by the presence of at least two cell populations with different genotypes in her retina, (II) the second mutation could have occurred in her retina as a somatic event, or (III) the wild type allele could have been silenced via epigenetic changes. The patient was negative to known X-linked RP pathogenic variants, which suggested a somatic mutation, mosaicism, or gene silencing as possible etiologies. This was in agreement with previous reports of unilateral RP.16,19,32 In order to better assess the distribution and potential role of her genetic variants in RP, we recommended extending genetic testing to any family member affected by retinal disease.

A recent study involving over two thousand patients with inherited retinal dystrophy found that a molecular diagnosis can be achieved via genetic testing in approximately 70% of patients.33 This leaves up to 30% of all retinal dystrophy cases without a definitive molecular diagnosis. Thus, although genetic testing is
informative, further research is needed to obtain the scientific and clinical knowledge to determine the role of novel mutations, like the ones identified in our patient. In this regard, the possibility of a novel AGBL5 and TULP/digenic form of RP cannot be ruled out completely. However, the unilateral presentation in our patient suggested a retina-specific event (e.g., somatic or epigenetic change) as a likely second hit. Again, to the best of our knowledge, this is the first report of a patient with probable URP expressing these two genetic variants in heterozygosity. Genetic tests for Inherited Retinal Diseases (IRD), like the one used for our patient, use blood or saliva samples to find genetic changes present in germline cells. Somatic mutations, low level mosaicism, or epigenetic changes fail to be detected on these tests. Of note, these have been proposed as mechanisms driving URP. To find the underlying molecular mechanism that would provide a definitive diagnosis for the patient, a biopsy of the retinal tissues would be needed.

Based on the clinical findings, her case history, and our quantitative data combined with her genetic finding of recessive variants expressed in heterozygosity, we propose either low level mosaicism or a somatic event involving her OS as a plausible explanation. Her genetic and clinical information has been included in the My Retina Tracker database and is now available to clinicians and researchers. Building a community of clinical scientists will enable us to better correlate genotypes and clinical manifestations in inherited retinal disease and to improve patient care.

Other clinical tests performed were useful for understanding and addressing the functional needs of our patient. It is our goal as low vision rehabilitation clinicians to enable our patients to function in their environment, maintain independence, and optimize the quality of their lives. Generally, most patients with only one severely impacted eye do not require low vision services. However, our patient, despite having a functionally normal OD, had both contrast loss OU and peripheral field loss OS that affected her bilateral visual function and for which management of these complaints was within the scope of low vision services. She also had the coexistence of early age-related macular degeneration in the macula of the OD more than the OS. Therefore, we felt it was also relevant to educate her on available resources should there be an eventual progression of macular degeneration.

Contrast sensitivity testing is an alternate method of quantifying the visual function of an individual. Often contrast sensitivity testing can detect a reduction in visual function earlier than visual acuity loss and can explain visual complaints that are disproportionate to acuity loss alone. In this case, our patient was experiencing severe contrast sensitivity reduction OS and mild to moderate contrast sensitivity loss OD. We feel this was due to the comorbidity of her macular
degeneration with RP. Enhancement of contrast sensitivity with yellow filters is a known treatment option and, in this case, provided our patient with some subjective improvement in her vision.\textsuperscript{34} In addition, the contrast enhancement feature of electronic video magnifiers and digital media, good lighting, and glare filters can be helpful for both URP and age-related macular degeneration.\textsuperscript{35-37}

The 120 point screening visual field and 30-2 threshold visual fields were used to assess the patient’s overall functional visual field, both in extent and sensitivity. A reverse telescope is ordinarily useful to enhance bilateral constriction of visual field; however this was not indicated or useful for our patient due to her better functioning OD. Orientation and mobility training can also be recommended for those with bilaterally reduced peripheral field loss. This training is ideally implemented prior to symptomatic vision loss to better prepare them for significant functional vision loss.\textsuperscript{37-39} For our patient, incorporating scanning techniques to her left side to compensate for the reduced left field was encouraged.

There are other functional considerations for patients with RP and macular degeneration. Driving rehabilitation may assist our patients to drive more comfortably and safely. Every region has its own regulations regarding driving requirements and recommendations. For those who no longer meet the requirements for driving in their jurisdiction, low vision rehabilitation can address alternate transportation options.\textsuperscript{40} For school aged children, they may receive supervision from Teachers of the Visually Impaired (TVI) and be provided with Individualized Education Programs (IEP).\textsuperscript{41,42} For those seeking employment, or currently employed, vocational training or rehabilitative services can be an option.\textsuperscript{35,40,41} For the elderly, independent living skills can assist them to better adapt to their home with the remaining vision.\textsuperscript{36} With reduced vision, individuals are more at risk to develop depression, so it is important to provide counseling services and encourage support from family members.\textsuperscript{36,43} A visual impairment can alter quality of life, but it should not prevent anyone from experiencing an independent fulfilling life.

**CONCLUSION**

Unilateral RP is a rare presentation of RP. When such a case presents itself, the clinician must thoroughly evaluate possible mimicking of RP. The literature on URP is scarce, thus more research is needed to fully understand this condition. It is important to evaluate the patient’s visual function as a whole, and to consider education and rehabilitation resources when appropriate to improve visual function. This case demonstrated that while the complications from her RP OS presented with peripheral vision issues, when combined with bilateral macular degeneration,
functional impairment occurred. Early intervention with low vision education was recommended and will aid the patient if vision continues to change from her comorbidities.

**Conflict of interests**

The authors have no financial or other relationships that might lead to conflict of interest.

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