Coexistence of Retinis Pigmentosa and Posterior Staphyloma: Case Series

Retinitis Pigmentosa ve Posterior Stefilom Birlikteliği: Olgu Serisi

Hasan KIZILTOPRAK[1], Bayram GÜLPAMUK[2], Kemal TEKİN[3], Mehmet Yasin TEKE[4]

ABSTRACT

We aimed to report a case series of coexistence of retinitis pigmentosa (RP) and posterior staphyloma (PS) and to describe their clinical outcomes. This case series include three patients' complete ophthalmological examinations including optical coherence tomography (OCT), fundus autoflorescence, fundus fluorescein angiography and enhanced depth OCT. All patients had myopic refraction and decreased visual acuity. Dilated fundus examination revealed waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentations, and large posterior staphyloma. Additionally, macular pseudohole (MPH) and cystoid macular edema (CME) were also detected. Coexistence of RP and PS is a rare condition that may result in serious visual complications. RP can also be accompanied by MPH and CME. As the surgical outcomes are not always satisfactory, vitreous surgery should be undertaken after careful consideration of the potential risks and benefits for such macular abnormalities.

Keywords: Cystoid macular edema, macular pseudohole, posterior staphyloma, retinitis pigmentosa.

ÖZ

Bu makalemizde, retinitis pigmentosa (RP) ve posterior stafilom (PS) birlikteliğini içeren bir vaka serisini sunmayı ve bunların klinik sonuçlarını tanımlamayı amaçladık. Bu vaka serisi, üç hastanın optik koherens tomografi (OCT), fundus otofloresansı, fundus flöresein anjiyografi ve EDİ-OCT'yi içeren tam oftalmolojik muayenelerini içermektedir. Tüm hastalarda miyopik refraksiyon ile görme keskinliğinde azalma mevcuttu. Dilate fundus muayenesinde balmumu optik atrofi, arteriyel atenüasyon, midperiferal kemik spikülü pigmentasyonlar ve büyük posterior stafilom saptandı. Ek olarak, maküler psödohole (MPH) ve kistoid maküler ödem (KMÖ) de tespit edildi. RP ve PS'nin varlığı ciddi görsel komplikasyonlarla sonuçlanabilen nadir bir durumdur. MPH ve KMÖ de eşlik edebilir. Cerrahi sonuçlar her zaman tatmin edici olmadığından, bu tip maküler anormallikler için vitreus cerrahisi dikkatle düşünülmelidir.

Anahtar Sözcükler: Kistoid maküla ödemi, maküler pseudohole, posterior stafiloma, retinitis pigmentosa.

INTRODUCTION

Retinitis pigmentosa (RP), characterized by rod and cone photoreceptor degeneration and progressive loss of peripheral and central vision, consists of a group of inherited progressive retinal dystrophies.[1] Retinal degeneration firstly begins with loss of the rod photoreceptors leading to nyctalopia, subsequently followed by the involvement of the cone photoreceptors leading to severe vision loss.[1] Both rod and cone function may be severely impaired or undetected at the final stage of the disease.[1]

Posterior staphyloma (PS) is a scleral ectasia from posterior expansion of eyeball and usually associated with pathological myopia. However, it has also been described in eyes without significant myopia.[2] Herein, we report a critical case series about the coexistence of RP and PS and also describe their clinical outcomes.

CASE 1

A 22-year old male patient presented with progressive nyctalopia and visual disturbance starting from the early

1- Asist. Dr., Ulucanlar Göz Eğitim ve Araştırma Hastanesi, Göz Hastalıkları, Ankara, Türkiye

2- Uz. Dr., Konya Beyhekim Devlet Hastanesi, Göz Hastalıkları, Konya, Türkiye

3- Uz. Dr., Kars Devlet Hastanesi, Göz Hastalıkları, Kars, Türkiye

4- Doç. Dr., Ulucanlar Göz Eğitim ve Araştırma Hastanesi, Göz Hastalıkları, Ankara, Türkiye

Geliş Tarihi - Received: 24.04.2018
Kabul Tarihi - Accepted: 08.05.2018
Ret-Vit 2019; 28: 198-203

Yazışma Adresi / Correspondence Adress:
Hasan KIZILTOPRAK
Ulucanlar Göz Eğitim ve Araştırma Hastanesi, Göz Hastalıkları, Ankara, Türkiye

Phone: +90 538 675 6528
E-mail: hsnkzltprk21@gmail.com

childhood. He had no history of systemic disorders, ocular trauma or surgery. Cycloplegic refraction was -6.00 (-1.25 x 175) diopter (D) in OD and -5.00 (-1.50 x 165) D in OS. The best corrected visual acuity (BCVA) was 20/63 in the right eye and 20/40 in the left eye. Intraocular pressures were within normal limits in both eyes. Examination of the anterior segment was unremarkable in both eyes. Slit lamp examination revealed bilateral vitreous degeneration. Fundus examination revealed waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentations, and large posterior staphyloma in both eyes with macular pseudohole (MPH) formation in the right eye (Figure 1). Spectral domain optic coherence tomography (SD-OCT) revealed progressive atrophy of retina and posterior staphyloma in both eyes and a MPH formation in the right eye (Figure 2). Additionally, fundus autoflorescence (FAF) imaging showed areas of hypoautofluorescence that were compatible with retina pigment epithelium (RPE) atrophy and fovea are seen as hyperautoflourescent area surrounded by a hypoautofluorescent ring in the right eye (Figure 3). Patient was diagnosed as coexistence of RP, PS, and MPH and no treatment was performed.

CASE 2

A 25-year old female patient presented with nyctalopia. She had no history of systemic disease, ocular trauma or surgery. Cycloplegic refraction was -7.00 (-1.00 x 10) D in OD and -7.00 (-1.50 x 105) D in OS. The BCVAs were 20/63 and intraocular pressures were within normal limits for both eyes. Anterior segment examinations were bilaterally unremarkable. Fundus examination revealed waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentations, and large posterior staphyloma in both eyes (Figure 4). SD-OCT revealed cystoid macular edema (CME) and concave shaped macula in both eyes (Figure 5).

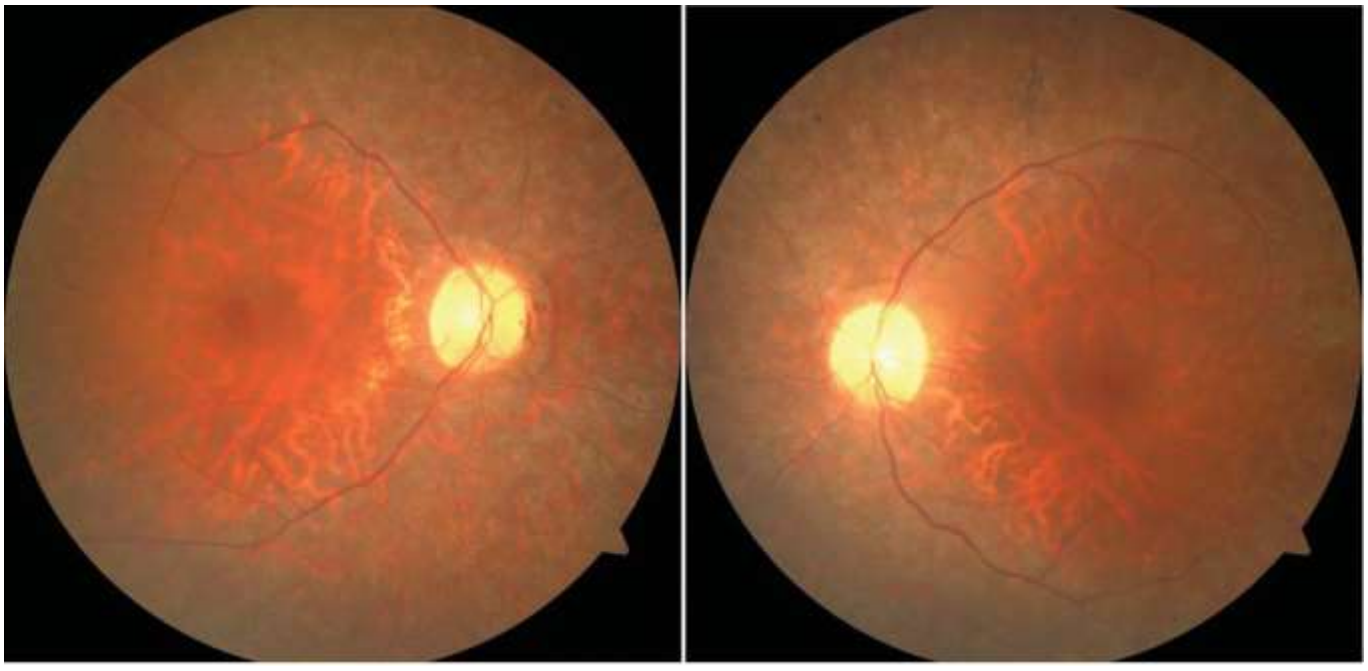

Figure 1. *Colored fundus images reveal waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentations, and large posterior staphyloma in both eyes with macular pseudohole formation in the right eye.*

Figure 2. *SD-OCT scans demonstrate progressive atrophy of retina and posterior staphyloma in both eyes and a MPH formation in the right eye.*

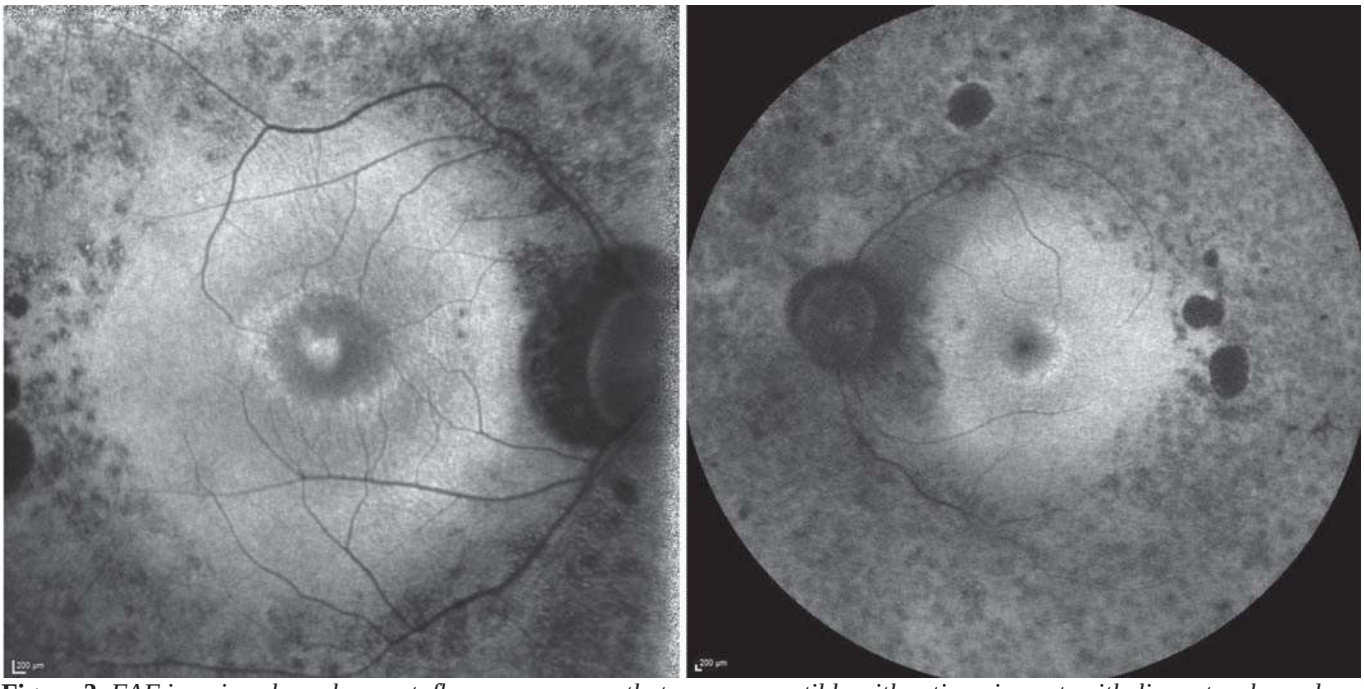

Figure 3. *FAF imaging shows hypoautoflorescence areas that were compatible with retina pigment epithelium atrophy and fovea is seen as hyperautoflorescent area surrounded by a hypoautofluorescent ring in the right eye.*

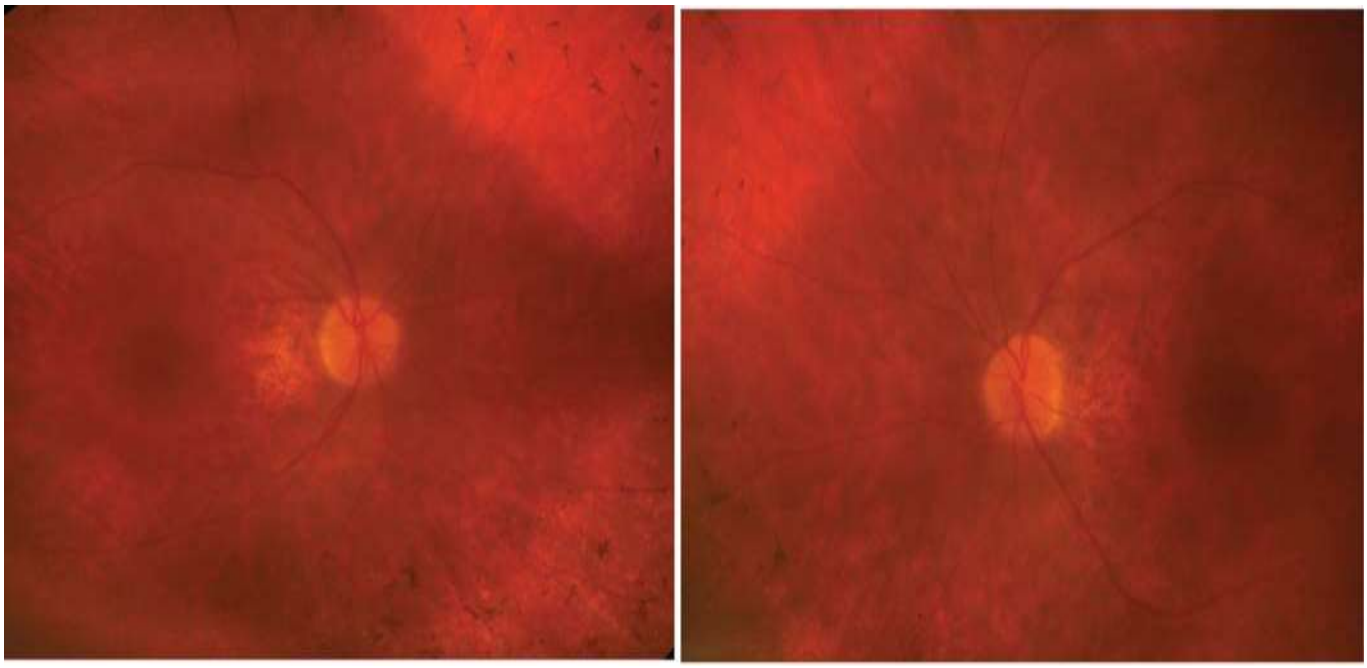

Figure 4. *Colored fundus images reveal waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentations, and large posterior staphyloma in both eyes.*

CASE 3

A 26-year old male presented with progressive decreased vision and nyctalopia. He had no history of systemic disease, ocular trauma or surgery. Cycloplegic refraction was -4.00 D in OD and -4.00 D in OS. The BCVAs were 20/40 and intraocular pressures were within normal limits for both eyes. Anterior segment examinations were unremarkable in both eyes. Fundus examinations revealed waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentation, and large posterior staphyloma in both eyes (Figure 6). Fundus fluorescein angiography revealed hypoflorescence in late phases (Figure 7). SD-OCT demonstrated posterior staphyloma and the choroidal thicknesses were within normal limits in both eyes with enhanced-depth OCT (Figure 8).

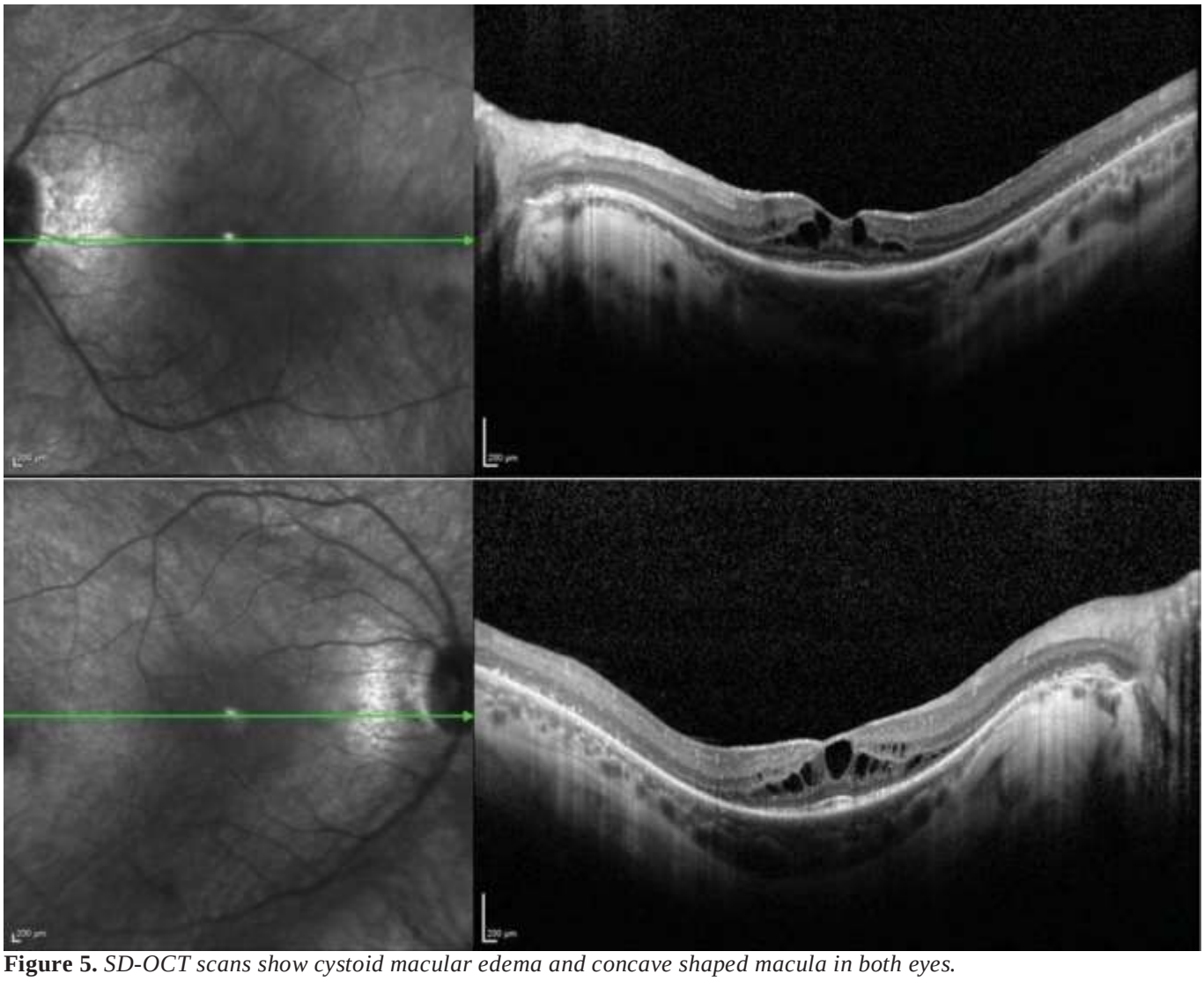

Figure 5. *SD-OCT scans show cystoid macular edema and concave shaped macula in both eyes.*

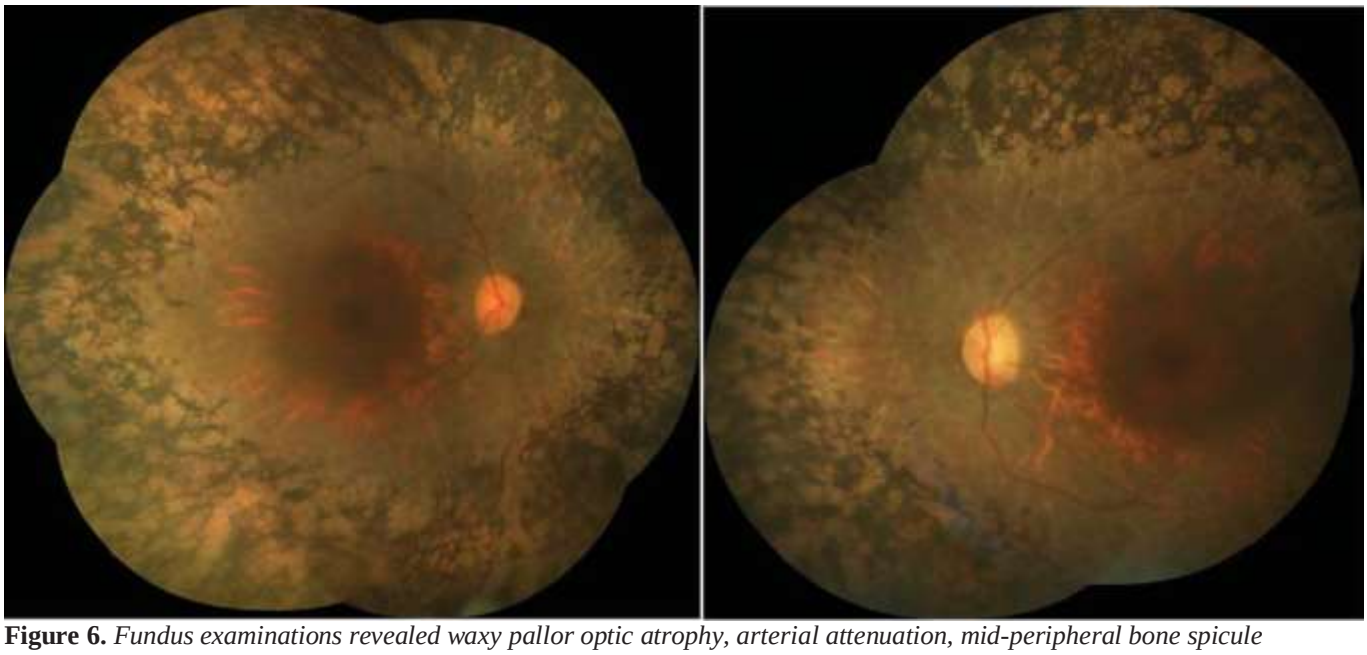

Figure 6. *Fundus examinations revealed waxy pallor optic atrophy, arterial attenuation, mid-peripheral bone spicule pigmentation, and large posterior staphyloma in both eyes.*

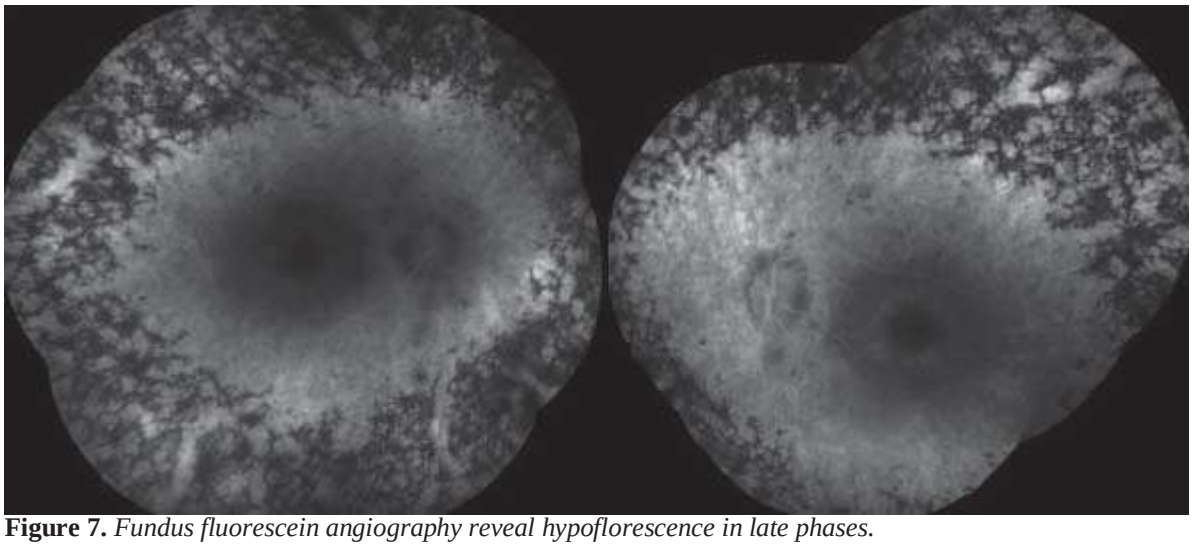

Figure 7. *Fundus fluorescein angiography reveal hypoflorescence in late phases.*

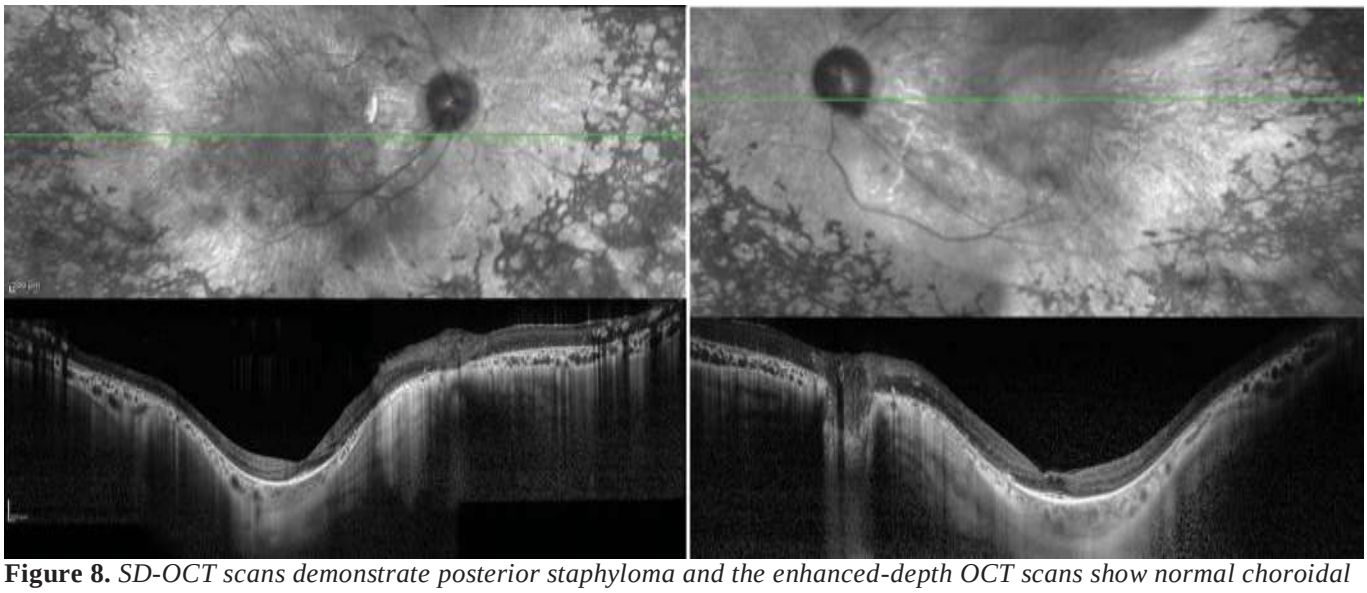

Figure 8. *SD-OCT scans demonstrate posterior staphyloma and the enhanced-depth OCT scans show normal choroidal thickness.*

DISCUSSION

RP is genetically heterogeneous, and clinical findings of the disease, the time of onset of symptomatic visual loss and the age at diagnosis vary. RP has a classical triad of waxy optic disc, arteriolar narrowing and bone spicule pigmentation in the retina and the diagnosis of RP is supported by full-field flash electroretinogram (ERG).[3] Patients generally complain about night blindness and progressive reduction in visual acuity. RP can be complicated by cataract, CME and visual field constriction.[4] Patients with RP also have vitreous degeneration; including vitreous gel retraction and posterior vitreous detachment. These changes may cause macular complications related to the vitreoretinal interface such as CME, macular hole, epiretinal membrane and vitreomacular traction syndrome.[5]

PS is an ectasia usually seen in high myopic patients and arising from localized protruding and thinning of the sclera. Various fundoscopic changes and complications including diffuse chorioretinal atrophy, patchy atrophy, RPE defects, lacquer cracks can develop in PS. There is an increased risk of visual loss, macular choroidal neovascularization, macular retinoschisis and macular hole formation.[6]

MPH has a very similar appearance to the lamellar macular hole and they must be reinterpreted on the basis of OCT findings. On FAF, there is a foveal tissue loss in true lamellar macular holes. Management of MPHs is observation unless there is a progressive visual decline that warrants surgery.[7]

RP, PS and MPH may separately result in severe visual impairment. Coexistence of these three entities can cause serious visual complications. Coexistence of RP and PS

has been previously described by Ilhan and colleagues.[8] We defined this coexistence in our three patients. In addition; we detected MPH and CME in our patients.

A rare dominantly inherited syndrome (MRCS) comprising microcornea, retinal dystrophy, cataract and posterior staphyloma has been reported previously in the literature.[9] A disease-causing mutation has also been subsequently described in the gene VMD2 in the MRCS pedigree. None of our patients had either microcornea or cataract. Therefore; we didn't need further genetic evaluations for these three patients.

RP is a progressive retinal degeneration and can be accompanied by high myopia. The elongation of eye can result in PS that can develop various fundoscopic changes like choroidal neovascularization, retinoschisis and epiretinal membrane formation. Contraction of an epiretinal membrane may lead to MPH or CME. There are several treatment options. In our first patient, no surgical intervention was planned, since both eyes had similar BCVA and the patient did not report any visual decline.

In conclusion, coexistence of RP and PS is a rare condition that may result in serious visual complications and can also be accompanied by MPH and CME. As the surgical outcomes are not always satisfactory, vitreous surgery should be undertaken after careful consideration of the potential risks and benefits for such macular abnormalities. Diagnosis is important not only for close monitoring of patients but also for the determination of such complications at early stages.

REFERENCES / KAYNAKLAR

1. Kalloniatis M, Fletcher EL. Retinitis pigmentosa: understanding the clinical presentation, mechanisms and treatment options. Clin Exp Optom 2004;87:65-80.
2. Mehta P, Dinakaran S, Squirrell D et al. Retinal pigment epithelial changes and choroidal neovascularisation at the edge of posterior staphylomas; a case series and review of the literature. Eye (Lond) 2004;20:150-3.
3. Na KH, Kim HJ, Kim KH et al. Prevalence, Age at Diagnosis, Mortality, and Cause of Death in Retinitis Pigmentosa in Korea-A Nationwide Population-based Study. Am J Ophthalmol 2017;176:157-65.
4. Strong S, Liew G, Michaelides M. Retinitis pigmentosa-associated cystoid macular oedema: pathogenesis and avenues of intervention. Br J Ophthalmol 2017;101:31-7.
5. Hagiwara A, Yamamoto S, Ogata K et al. Macular abnormalities in patients with retinitis pigmentosa: prevalence on OCT examination and outcomes of vitreoretinal surgery. Acta Ophthalmol 2011;89:122-5.
6. Saka N, Ohno-Matsui K, Shimada N et al. Long-term changes in axial length in adult eyes with pathologic myopia. Am J Ophthalmol 2010;150:562-8.
7. Gaudric A, Aloulou Y, Tadayoni R et al. Macular pseudoholes with lamellar cleavage of their edge remain pseudoholes. Am J Ophthalmol 2013;155:733-42.
8. Ilhan A, Yolcu U, Diner O et al. Coexistence of Posterior Staphyloma, Retinitis Pigmentosa and Moderate Myopia. J Coll Physicians Surg Pak 2016;26:70-1.
9. Michaelides M, Urquhart J, Holder GE et al. Evidence of genetic heterogeneity in MRCS (microcornea, rod-cone dystrophy, cataract, and posterior staphyloma) syndrome. Am J Ophthalmol 2006;141:418-20.