Posterior Microphthalmos: Assessment with Clinical and Imaging Features of 3 Cases

Posterior Mikroftalmi: 3 Vakanın Klinik ve Görüntüleme Bulgularıyla Değerlendirilmesi

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ABSTRACT

Posterior microphthalmos (PM) is a relatively rare type of microphthalmos in which the posterior segment is predominantly affected with normal anterior segment findings. The main findings are high hypermetropia and elevated papillomacular folds. This paper consists of clinical and imaging findings of 3 PM cases. All cases had high hypermetropia (+17.00, +19.00, +17.00 dioptry) in cycloplegic refraction. The dilated fundus examination revealed elevated papillomacular folds in all the cases and crowded disk in only 2 of the 3 cases. In the B-scan ultrasound evaluation sclerochoroidal thickening, decreased total axial length (14.6-15.0 mm), and marked foreshortening of the posterior segment were noted. Fundus fluorescein angiography was performed in 2 patients and in those in which angiography was performed the foveal avascular zone seemed to be significantly reduced. Optic coherence tomography demonstrated the elevated papillomacular folds seen on dilated fundus examinations. The diagnosis of PM should be considered in patients with high axial hypermetropia, elevated papillomacular retinal folds, and normal anterior segment examination. Patients should be followed up properly for refractive amblyopia and for the complications that may emerge.

Key Words: Posterior microphthalmos, hypermetropia, papillomacular fold.

ÖZ

Posterior mikroftalmi (PM) normal ön segment bulgularıyla birlikte arka segmentin baskın olarak tutulduğu nadir bir mikroftalmi şeklidir. Temel bulguları yüksek hipermetropi ve yüzeyden kabarık papillomaküler retinal katlantılardır. Bu yazı 3 PM olgusunun klinik ve görüntüleme bulgularını içermektedir. Bütün olgularda sikloplejik refraksiyonda yüksek hipermetropi (+17.00, +19.00, +17.00 dioptri) mevcuttu. Dilate fundus muayenesinde tüm hastalarda yüzeyden kabarık papillomaküler katlantılar ve 3 hastanın 2 sinde kalabalık optik sinir başı görüldü. B-mod ultrason değerlendirmesinde sklerokoroidal kalınlaşma, azalmış total aksiyel uzunluk (14.6-15.0 mm) ve arka segmentin belirgin kısalığı izlendi. Fundus floresein anjiografi 2 olguda yapıldı, yapılan hastalarda foveal avasküler alanda ciddi daralma görüldü. Optik koherens tomografi dilate fundus muayenesinde izlenen yüzeyden kabarık papillomaküler foldları göstermiştir. PM tanısı aksiyel hipermetropi, yüzeyden kabarık papillomaküler retinal katlantılar ve normal ön segment muayenesi olan hastalarda düşünülmelidir. Hastalar refraktif ambliyopi ve gelişebilecek komplikasyonlar açısından doğru şekilde takip edilmelidir.

Anahtar Kelimeler: Posterior mikroftalmi, hipermetropi, paillomaküler katlantı.

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Geliş Tarihi - Received: 21.09.2011
Kabul Tarihi - Accepted: 28.12.2011
Ret-Vit 2012;20:141-145

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INTRODUCTION

Microphthalmos is defined as a developmental condition in which the axial length of the eye is smaller by more than two standard deviations when compared to age-matched controls.[1,2]

It is subclassified as simple or complex. Simple microphthalmos is a congenitally small eye without major ocular findings, while complex microphthalmos is the condition where ocular abnormalities coexist.[3,4]

In microphthalmic eye frequent findings were shorter anterior and posterior segment, high hypermetropia, and small cornea.[5]

Nanophthalmos is a kind of microphthalmos with microcornea, shallow anterior chamber, relatively high lens/globe index, and thick sclera having a tendency for spontaneous or surgical uveal effusion.[6]

Posterior microphthalmos (PM) is a relatively rare type of microphthalmos in which the posterior segment is predominantly affected with normal anterior segment findings. The main findings in this particular condition are high hypermetropia and elevated papillomacular folds.[2,7]

In this paper 3 PM cases are presented along with their fundus angiography (FA), optical coherence tomography (OCT), and B-scan ultrasound findings.

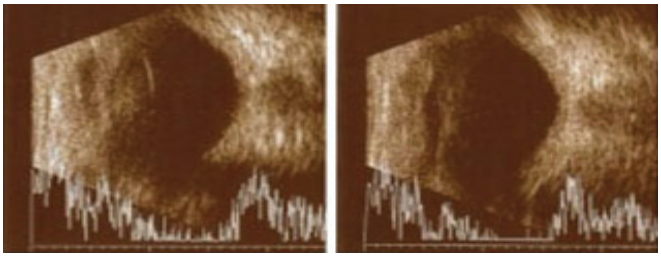

Figure 1: *B-scan ultrasound of Case 1. Increased sclerochoroidal thickness and shallow vitreous cavity are seen in both eyes.*

CASE REPORT

Case 1

A 12-year-old female patient having best corrected visual acuities (BCVA) of 0.1 and 0.2 in the right and left eye respectively on the Snellen chart was referred to our retina department. The patient was the only child of non-intermarriage and had a history of glasses wear since she was 5 years old.

Systemic, mental, and external ophthalmologic examinations were found to be completely normal. Cycloplegic refractometry findings were +17.00 (+0.50x90) diopters for both eyes.

Total axial length measurements were 14.9 mm for the right eye and 14.4 mm for the left eye.

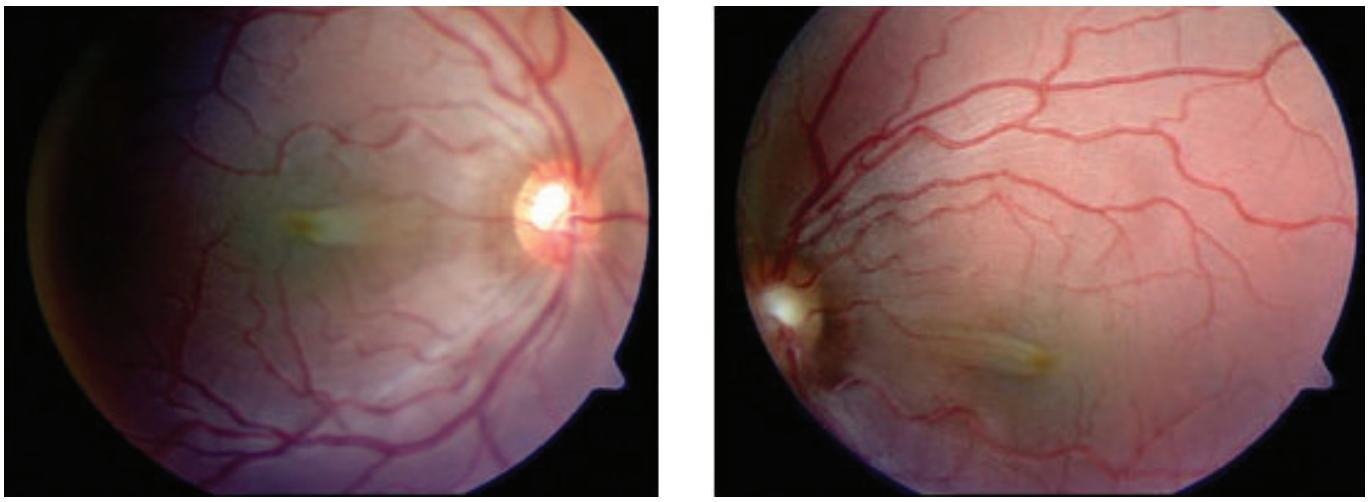

Figure 2: *Color fundus image of Case 1. See the infero-oblique displacement of the macula and papillomacular retinal fold in both eyes.*

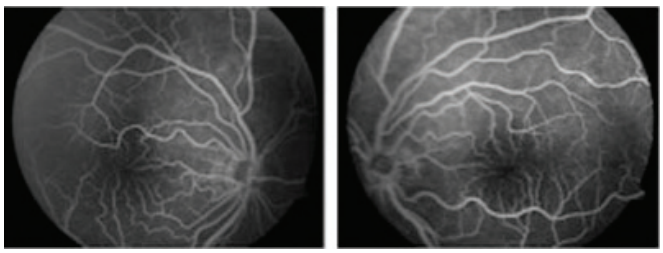

Figure 3: *FA of Case 1. See the severely reduced foveal avascular zone in both eyes.*

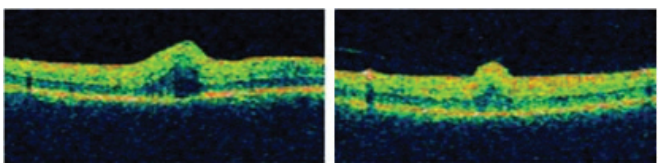

Figure 4: *OCT images of Case 1. Elevated papillomacular folds are seen with a crowded neurosensory retina.*

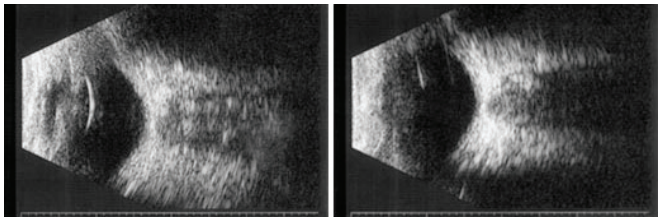

Figure 5: *B-scan ultrasound of Case 2. Both eyes have increased sclerochoroidal thickness and significant decrease in vitreous depth.*

The B-scan ultrasound evaluation revealed decreased length of vitreous cavity and total axial length along with sclerochoroidal thickening (Figure 1).

The anterior segment and angle structures were normal. The dilated fundus examination revealed a slightly elevated oblique papillomacular retinal fold, slight inferior oblique displacement of macula, and normal optic nerve head. No retinal pigmentary changes were encountered (Figure 2). The fundus angiography examination was normal other than a reduced foveal avascular zone (FAZ) (Figure 3). The OCT showed retinal folds especially prominent in the papillomacular area, which was consistent with fundus findings (Figure 4).

Case 2

An 11-year-old male patient with a BCVA of 0.3 and 0.2 on the Snellen chart in the right and left eye respectively with a history of glasses wear since 3 years old was examined in the retina department. Systemic and mental examinations showed no abnormality. His two brothers' and parents' ophthalmologic examinations were normal. Cycloplegic refractions were +19.00 (+1.00x70) diopters in the right eye and +18.00 diopters in the left eye.

The B-scan ultrasound evaluation revealed total axial length measurements as 15 mm in the right eye and 14.9 mm in the left eye with a shallow vitreous cavity and increment in sclerochoroidal thickness (Figure 5). Having a normal anterior segment on dilated fundus examination we found an inferiorly displaced macula, elevated papillomacular folds, and crowding of the optic nerve head (Figure 6). FAZ was found to be markedly reduced in FA.

We were not able to obtain clear images in FA due to the patient's lack of cooperation and so we did not include them in the paper. Elevated papillomacular folds compatible with fundus findings were seen in OCT images (Figure 7).

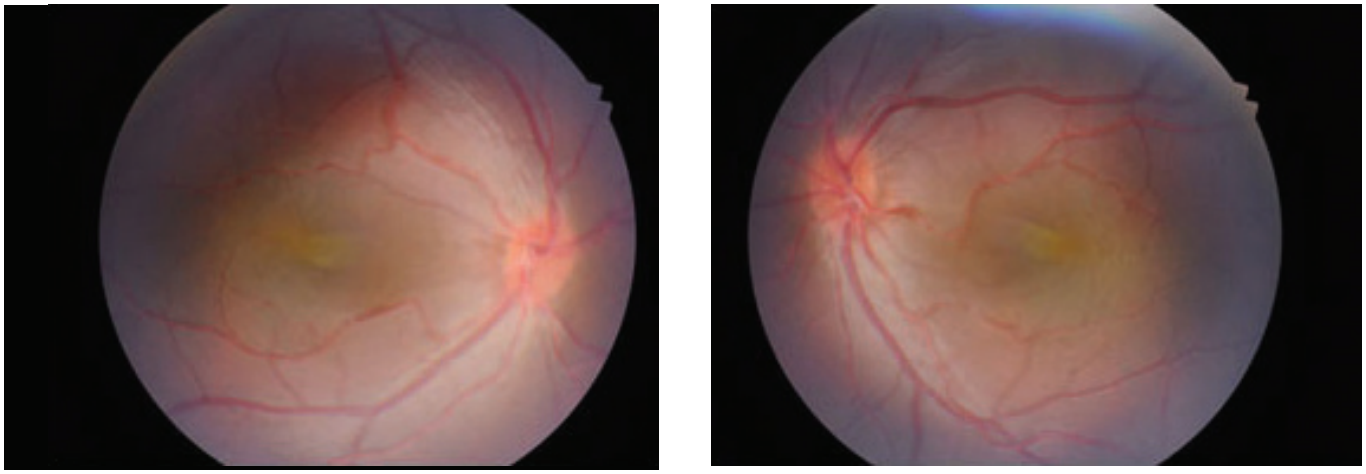

Figure 6: *Color fundus images of Case 2. Papillomacular retinal folds and slight infero-obliquely displaced macula.*

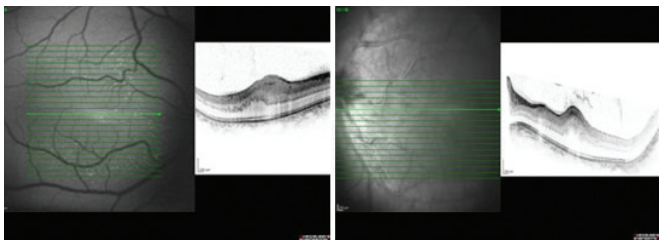

Figure 7: *OCT images of Case 2. Papillomacular folds are evident in both eyes with a relative crowded neurosensory retina in the right eye.*

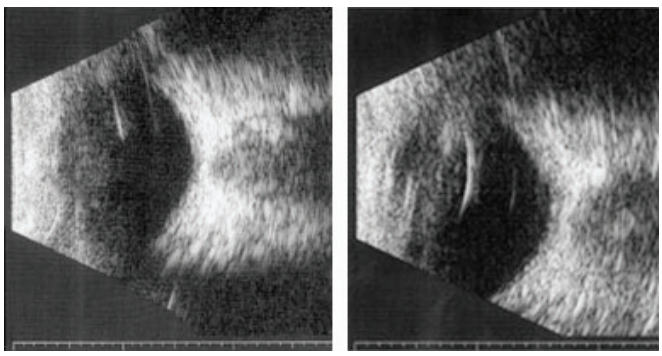

Figure 8: *B-scan ultrasound of Case 2. Decreased vitreous cavity length causing the decrease in total axial length of both eyes.*

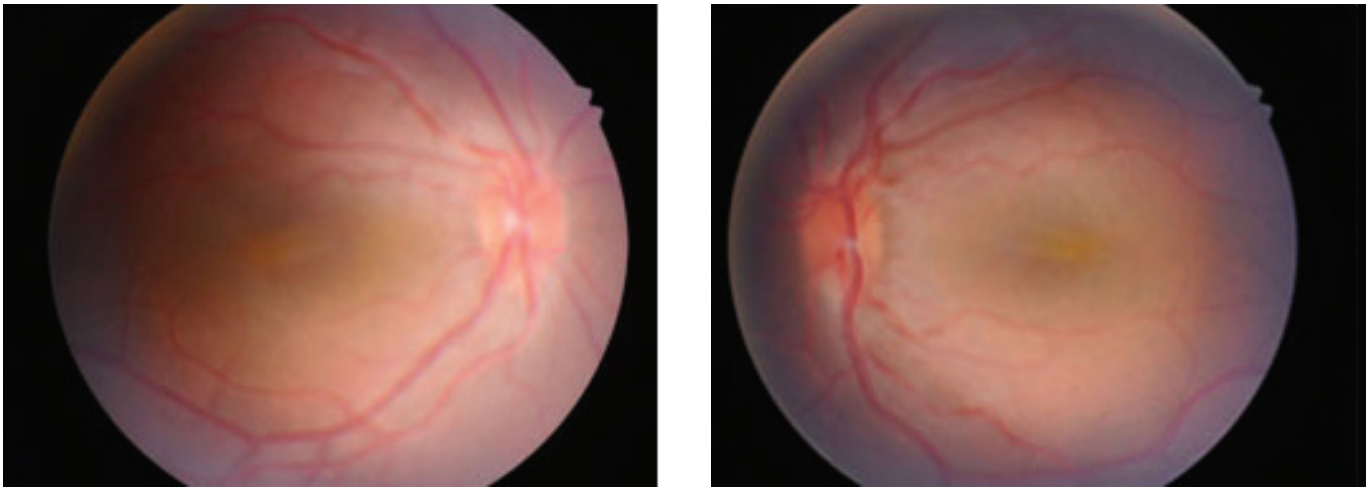

Figure 9: *Color fundus images of Case 3. See the papillomacular folds in both eyes and infero-oblique displacement of the macula in the right eye.*

Case 3

An 8-year-old female patient having 0.2 BCVA on the Snellen chart for both eyes with a history of glasses wear since 5 years old was evaluated in the retina department. Her parents' and sister's ophthalmologic examinations were normal. The result of cycloplegic refractions was +17.00 diopters for both eyes. Total axial length measurements on the B-scan ultrasound examination were 14.7 mm and 14.6 mm for the right and left eye respectively along with shallow posterior segment and sclerochoroidal thickening (Figure 8). In both eyes there were elevated oblique papillomacular folds and crowding of the optic nerve head on dilated fundus examinations (Figure 9).

We were unable to obtain consent from the parents for FA and so we lack FA images for this patient. The elevated papillomacular folds seen in the fundus examination were also documented in the OCT evaluation (Figure 10).

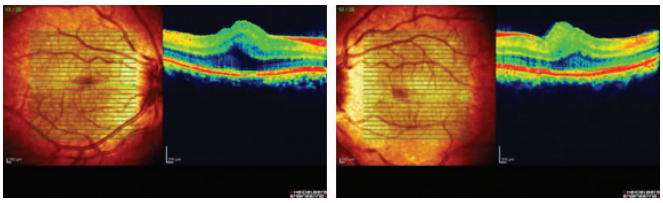

Figure 10: *OCT images of Case 3. Papillomacular folds compatible with dilated fundus findings are seen.*

DISCUSSION

The condition in which posterior segment development is disproportionately affected with normal anterior segment structures is named PM and is classified in the simple microphthalmos.[7] It is characterized by foreshortening of the posterior segment and normal corneal diameters, anterior chamber depth, and lens thickness.[9] The main findings are high hypermetropia and elevated papillomacular folds.[8]

Authors have suggested that abnormal sclera in PM failed to grow normally and as the sensory retina continues to grow normally the disproportion between the growth of sclera and retina caused the sensory retina to fold and resulted in typical papillomacular retinal folds and high hypermetropia due to decreased total axial length.[2,10]

The disease is thought to have an autosomal recessive inheritance and the membrane-type frizzled-related protein (MFRP) is the only gene so far reported to be responsible.[11] A study conducted on Tunisian families isolated a novel locus for non-syndromic posterior microphthalmos on chromosome 2q37.1.

Gal et al.,[12] studied PM patients and isolated an abnormality with the expression of a proteolytic enzyme that is encoded by PRSS56. Abnormal proteolytic activity might cause the accumulation of the proteins in sclera, thus causing a thicker sclera, which limits the growth of posterior segments. This finding is of great importance in clarifying the assumed pathogenesis of the disease.

Other abnormalities that might be associated with this condition are the absence of FAZ, retinal striae, uveal effusion, pigmentary retinal dystrophy, pseudopapilledema, chorioretinal folds, and macular hole.[2,7]

All 3 of our patients had high hypermetropia (+17.00, +19.00, +17.00 diopters), oblique papillomacular retinal folds, and sclerochoroidal thickening in ultrasound examinations, along with normal appearing anterior segments, which led us to the diagnosis of PM. The main findings of the condition are high hypermetropia in all patients and retinal folds, which were seen in 72.2% of the patients.[2] Spitznas et al.,[7] who first described the condition, stated that FAZ was absent.

However, this finding was not confirmed by subsequent studies.[2,10] We think that it is more appropriate to regard this situation as absent or reduced FAZ as stated in the study by Khairallah et al.,[2] In our patients the FAZ was reduced in the 2 patients in whom we were able to perform FA.

In some of the studies the optic nerve head was reported to be crowded.[2,7,8] We observed the crowded disk phenomenon in cases 2 and 3 but not in case 1.

The sclerochoroidal thickening seen in all 3 patients was noted as a rare finding in some previous studies.[10,13] However, in the biggest review of PM, with 18 patients performed by Khairallah et al.,[2] thickening of sclera and choroid was seen in all patients and they referred to this phenomenon in the possible pathogenesis of the PM.

Pigmentary retinopathy might be associated with PM although not frequently as fundus albipunctatus.[7] Recently a new syndrome called Posterior Microphthalmos Pigmentary Retinopathy (PMPRS) was described, comprising PM, pigmentary retinopathy, optic nerve head drusen, and foveoschisis.[14] We did not see any pigmentary changes in our cases. In case 2, we think that the hyper-autofluorescence spots in fundus autofluorescence mode, not seen in the color fundus photo, were due to fundus albipunctatus accompanying PM.

Due to the specific abnormalities of the ophthalmic structures PM patients may experience some complications during their follow-up. Uveal effusion is one of these conditions and it may appear both spontaneously or related to a surgical intervention.[2]. Sometimes it may be necessary to examine the peripheral retina to see subtle uveal effusions and perform proper sclerotomies. Moreover, the surgical interventions should be performed by skillful surgeons who are aware of the disease and the conditions that they might face during the surgery. The most important problem in PM patients is ametropic amblyopia. An early diagnosis along with proper refractive examination and follow-up is crucial to avoid amblyopia.

In conclusion, PM is a disease characterized by high hypermetropia, elevated papillomacular folds, sclerochoroidal thickening, and shallow vitreous with a normal anterior segment. In patients with high hypermetropia and normal anterior segment evaluation, in the diagnoses of PM dilated fundus examination, ultrasound and OCT evaluation will be sufficient. Fundus angiography may be useful to detect some associated retinal pathologies.

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