

Original Article

Ocular findings in posterior microphthalmos



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Abstract

Aim: To report a critical case series of six patients with posterior microphthalmos (PM).

Method: Complete ophthalmologic examinations of all patients were performed using best-corrected visual acuity (BCVA), cycloplegic refraction, applanation tonometry, slit lamp biomicroscopy of the anterior segment, funduscopy, A and B mode ultrasonography (USG), keratometry, and optic coherence tomography (OCT).

Results: The most significant clinical characteristics of male patients aged 10–25 years was the presence of shorter posterior segments (mean: 15.27–18.91 mm) accompanying high hyperopia (mean +9.00 – +18.50 diopter) despite the normal anterior segment findings. The BCVA ranged between 20/320 and 40/100. Retinal folds were detected bilaterally on the papillomacular band in all patients. Although neurosensory retina was included in the fold in OCT images, retinal pigment epithelium, choroid, and sclera were not included in the fold. Pigmentary retinopathy was detected in one patient.

Conclusion: Despite normal anterior segment, posterior microphthalmos is characterized with high hyperopia, and shorter axial length and bilateral papillomacular retinal fold. Refractive amblyopia, uveal effusion syndrome, retinal detachment and macular hole are complications that can be corrected. Posterior microphthalmos must be kept in mind in patients with a normal anterior segment, and high hyperopia.

Keywords: High hyperopia, Retinal papillomacular fold, Posterior microphthalmos

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Introduction

The developmental disorder of microphthalmos is characterized by a total axial length of globe being at least two deviations below compared with the people of the same age group.¹ Posterior microphthalmos (PM) is a rare subtype of microphthalmos characterized by short axial length and a relatively normal anterior segment. The most frequent clinical findings are hyperopia and retinal papillomacular fold.^{2,3} Researchers reported other clinical findings that might accompany PM such as uveal effusion, macular hole, pigmentary retinopathy, retinoschisis, foveoschisis, and retinal

detachment in the literature.^{2–9} Although researchers reported that PM might have an autosomal recessive heritage, most reported cases were sporadic.^{2,4}

Material and methods

Six patients with posterior microphthalmos with high hyperopia and normal anterior segment imaging and significant short posterior segments were included in the study. Complete ophthalmologic examinations of all patients consisting of best-corrected visual acuity (BCVA), cycloplegic refraction, applanation tonometry, slit lamp biomicroscopy

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of the anterior segment, funduscopy, A and B mode ultrasonography (USG), keratometry and optic coherence tomography (OCT) was performed. Fundus pictures of all patients were taken, and fundus fluorescein angiography (FFA) was performed to 4 patients excluding two patients aged 10 and 11 years. This study was performed according to the tenets of Declaration of Helsinki and detailed information about the procedures was given to all patients and written and verbal informed consent were taken from them.

Results

The mean age of the patients was 16 years (range, 10–25 years). Although 4 patients were sporadic, 2 patients were siblings and their parents were consanguineous. One brother among 8 siblings who we could not examine was reported to have undiagnosed vision loss. No other health problems were detected in the patients. Ophthalmologic examinations of the parents were completely normal.

High hyperopia was detected in all patients. Cycloplegic examinations were performed 30 minutes after administration of 1% cyclopentolate eye drops. Cycloplegic refraction levels were calculated between +9.00 and +18.50 Diopters. BCVA according to Snellen charts ranged between 20/320 to and 40/100. The total axial length, lens thickness and anterior chamber depth were calculated using a scanning USG. Axial lengths below the normal values ranged between 15.27 and 18.91 mm (Table 1). Anterior segment findings were found normal in biomicroscopy examinations, and in A scan USG calculations. Anterior chamber depth and lens thickness were within normal ranges (range, 2.99–3.82, and 3.79–4.20 mm, respectively). Corneal curvature levels were calculated between 6.5 mm and 7.5 mm in keratometry (Table 2). Intraocular pressures were calculated using Goldmann applanation tonometry, and were found normal in all eyes. Although amblyopia was a common clinical finding, all patients were orthopic in the primary position.

A horizontally underlying papillomacular retinal fold from the center of fovea to the optic disc, and crowded optic disc were detected in all patients in dilated fundus examination (Fig. 1). Pigmentary retinopathy was detected in one patient. Papillomacular retinal folds compatible with fundus image were detected in OCT examination, and although

neurosensory retinas were included in the fold, retinal pigment epithelium, choroid, and sclera were not included (Figs. 2 and 3).

Discussion

Posterior microphthalmos, which is characterized by a normal anterior segment and decreased posterior segment length, is classified as simple microphthalmos. Franceschetti and Gernet first described the association of normal corneal diameter, and high hyperopia as posterior microphthalmos.¹⁰ Relhan et al. differentiated nanophthalmos (NO) and posterior microphthalmos considering the horizontal corneal diameter in their study published in 2015. Corneal diameter ≥ 11 mm was regarded as PM, and ≤ 11 mm was regarded as nanophthalmos.¹¹ The main clinical findings of posterior microphthalmos are high hyperopia and retinal folds.² Elevated papillomacular retinal folds compatible with fundus images are detected in OCT images. Involvement of neurosensory retina to folds, and exclusion of retinal pigment epithelium, choroid, and sclera to folds are also examined in OCT images.^{12,13}

Although the pathogenesis of posterior microphthalmos is controversial, the opinion of Bonton and Purnell is generally accepted who suggested that growth of the outer layer of the eye such as sclera, choroid, and retinal pigment epithelium is inhibited, whereas growth in the inferior layer of the sensorial retina continues, thus leading to a retinal fold, short total axial length, and high hyperopia.¹⁴

Researchers claimed the responsible protein as membrane frizzled-related protein (MFRP) in the etiopathogenesis of posterior microphthalmos, which was suggested to be inherited in an autosomal recessive way.¹⁵ Raul Ayala-Ramirez et al. described MFRP mutations in 4 siblings in a family in a syndrome with accompanying posterior microphthalmos, retinitis pigmentosa, foveoschisis, and optic nerve drusen.⁴ The pigmentary retinopathy finding mentioned in their study was detected in only one patient in our study. Noiwalay et al. detected PRSS56 and MFPR mutations in patients with posterior microphthalmos.¹⁶

High hyperopia, short axial length, and papillomacular retinal fold accompanying normal anterior segment findings were detected in all our patients. The layered convex papillomacular retinal folds, which are described as the most

Table 1. Demographic data and findings of ophthalmic examination.

Patient no	Age/Gender	BCV		Refraction (diopters)		Axial Length (mm)		Ophthalmoscopy
		Right	Left	Right	Left	Right	Left	
1	16/M	20/100	20/100	+11.75 + 050 × 140°	+13.25 + 1.50 × 30°	18.91	18.59	Horizontal papillomacular retinal fold
2	25/M	20/250	20/320	+16.50 + 1.25 × 160°	+15.75 + 1.25 × 175°	17.76	18.00	Horizontal papillomacular retinal fold
3	10/M	20/100	20/200	+18.25–0.75 × 59	+18.50– 0.25 × 100	15.52	15.27	Horizontal papillomacular retinal fold
4	11/M	20/320	20/200	+13.00	+13.00	16.20	16.10	Horizontal papillomacular retinal fold,
5	17/M	20/100	20/100	+17.00	+17.00	15.50	15.30	Horizontal papillomacular retinal fold and pigmentary retinopathy
6	13/F	40/100	40/100	+9.00 + 0.50 × 130	+9.25	18.31	18.50	Horizontal papillomacular retinal fold

Table 2. Anterior segment and intraocular pressure measurements of patients.

Patient no	Corneal curvature		Anterior chamber depth		Lens thickness		Intraocular pressure (mm Hg)	
	Right eye	Left eye	Right eye	Left eye	Right eye	Left eye	Right eye	Left eye
1	7.50	7.00	2.99	2.97	4.11	4.20	18	15
2	7.40	7.10	3.01	3.11	4.10	4.08	16	18
3	6.60	6.50	3.25	3.10	3.82	3.79	15	16
4	7.10	7.30	3.29	3.26	4.01	4.10	15	17
5	7.20	7.00	3.82	3.80	3.90	3.95	17	18
6	7.40	7.40	3.26	3.20	3.95	3.95	15	14

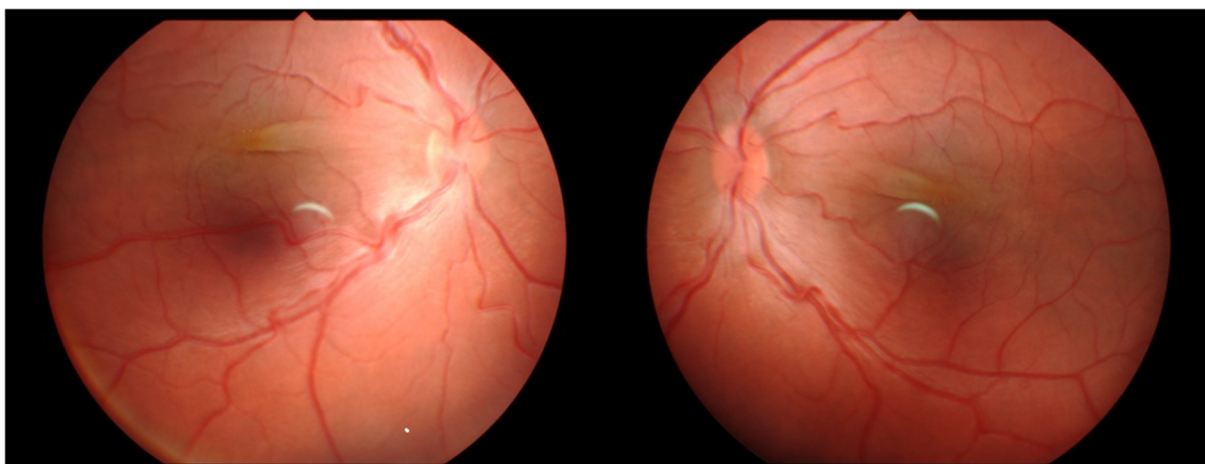


Fig. 1. Fundus images showing the papillomacular folds.

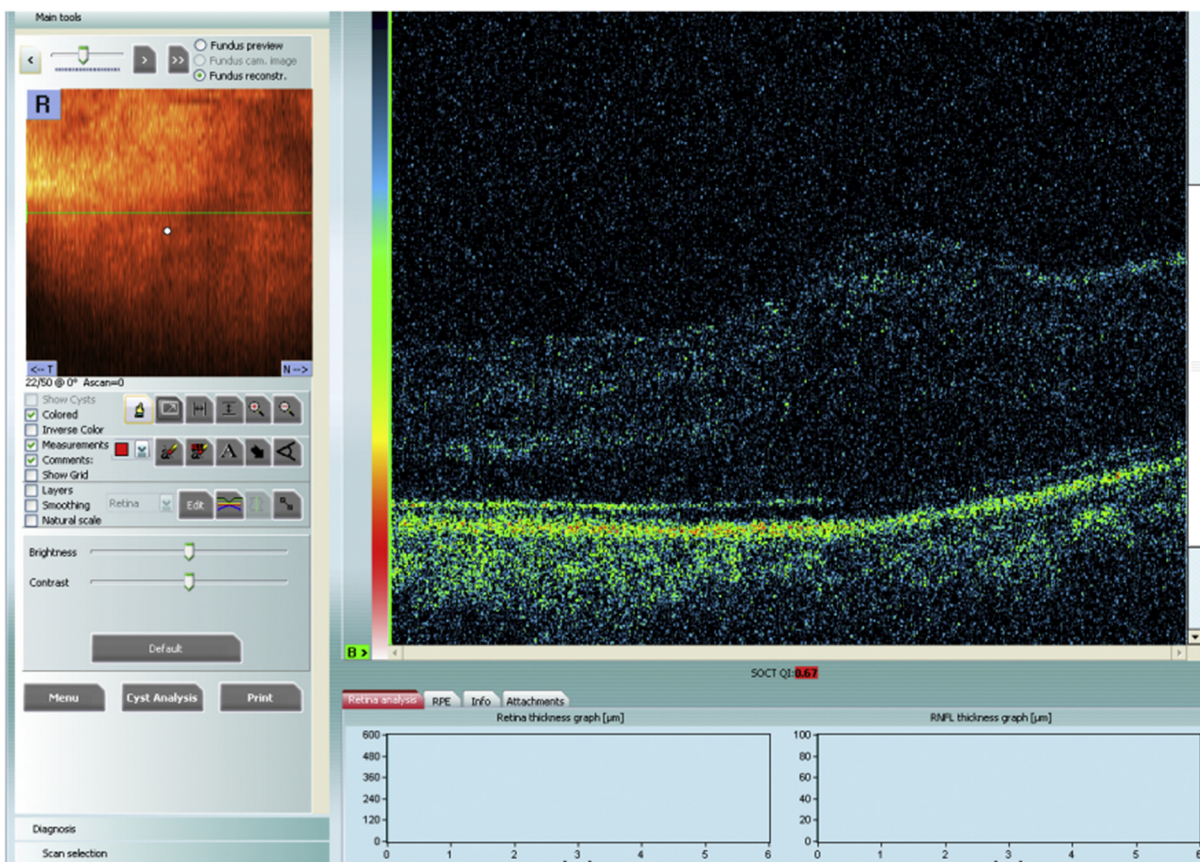


Fig. 2. Optical coherence tomography image of the macular fold without involvement of retinal pigment epithelium.

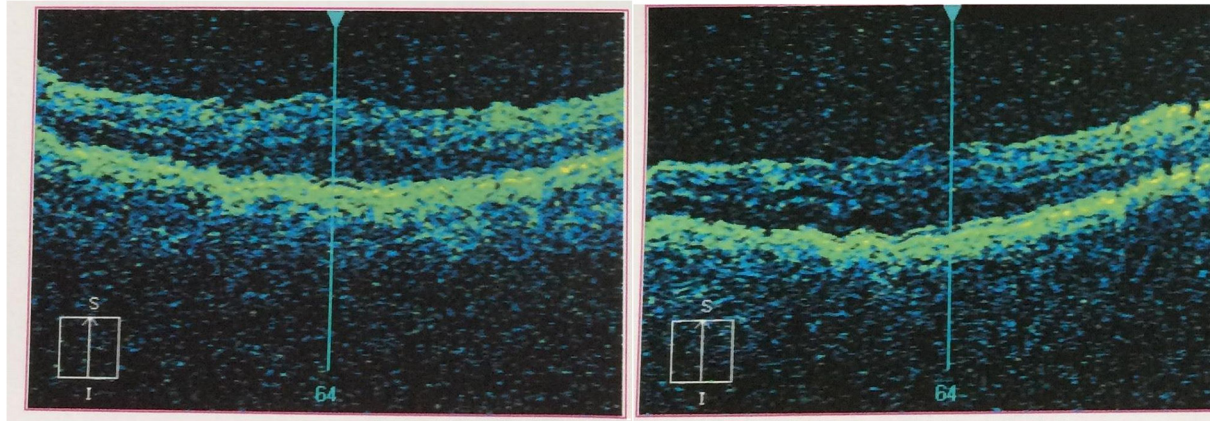


Fig. 3. Optical coherence tomography images of the papillomacular folds.

common finding of posterior microphthalmos, was detected with a prevalence of 72.2% in a series of 18 patients in a study by Khairallah et al.³ Although chorioretinal folds were detected in 11 patients in that study, we detected no chorioretinal folds, and they have rarely been reported in other studies.³ Tekin et al. detected papillomacular fold in all patients.¹³

Although Spitznas et al. claimed that a foveal avascular zone loss was detected in all patients with PM, it was rarely or never detected in other studies.^{2,3,14,17} Crowded optic nerve appearance was detected in all patients in our study. This finding was also reported in several studies.^{2,3,5,6}

Various complications may develop in the follow-up of patients with posterior microphthalmos. Uveal effusion is one of the complications that may develop spontaneously or due to surgical interventions.³ Attentive peripheral fundus examination and B-scan USG are valuable in diagnosis and follow-up of uveal effusion.³ Khairallah et al. in their case series with 18 patients reported uveal effusion syndrome in 3 patients, and closed angle glaucoma developed in one patient due to uveal effusion.³ However, no uveal effusion was detected in the follow-up of our patients, and intraocular pressures were calculated as normal.

Lee et al. reported bilateral macular holes in a case of posterior microphthalmos.⁷ Kiratlı et al. detected venous congestion and arteriolar narrowing, particularly in the peripheral temporal retina, in the fundus examination of a case of posterior microphthalmos with significant absent cyclotorsion of macula and temporal veins, and reported that the findings might eventually cause complications such as subretinal fluid, macular dragging, and retinal attachment.¹⁸ Park et al. in their series with 4 cases reported optic nerve hypoplasia and newly-developed neurosensory retinal detachment in one patient.⁸ Khairallah et al. detected pigmentary retinopathy in 4 patients.³

High hyperopia and associated amblyopia, papillomacular folds, and other possible complications are the most significant causes of vision loss in patients with posterior microphthalmos.³ The diagnosis may be overlooked owing to a normal anterior segment appearance. Therefore, recognition of posterior microphthalmos will help to treat the accompanying hyperopia and amblyopia, and to take precautionary measures against the complications. Early diag-

nosis must be performed using dilated fundus examination, biometrics, ocular ultrasonography, and optic coherence tomography.

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Declaration of interest

The authors declared that there is no conflict of interest.

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