Detailed Ophthalmologic Evaluation of Posterior Microphthalmos

Zeynep Alkin[1], Abdullah Ozkaya[1], Yalcin Karakucuk[1], Ahmet Demirok[1,2]

ABSTRACT

We performed various ophthalmic investigations in order to confirm the diagnosis and document the various features of posterior microphthalmos in a 21-year-old male. Ophthalmic examination revealed low vision with high hyperopia, papillomacular folds, midperipheral pigmentary changes and crowded optic discs. The optic discs were small and crowded with increased nerve fiber layer thickness. Fundus fluorescein angiography showed reduced diameter of a capillary free zone. Anterior segment (AS) optical coherence tomography demonstrated near normal anterior chamber depths, but markedly diminished anterior chamber angles. In spite of the increased corneal thickness and steep corneas, lens thickness and endothelial cell counts were normal. Sclerochoroidal thickening and foreshortening of the globes were detected with B-scan ultrasonography. Electroretinographic findings and visual field tests were similar to those in pigmentary retinopathy. Posterior microphthalmos is a complex eye disorder, which affects predominantly the posterior segment but also involves the AS of the eye.

Key words: Anterior Segment Optical Coherence Tomography, B-scan Ultrasonography, Fundus Autofluorescence, Fundus Fluorescein Angiography, Posterior Microphthalmos, Spectral Domain Optical Coherence Tomography

Access this article online

Website: www.meajo.org

DOI: 10.4103/0974-9233.129774

Quick Response Code:

INTRODUCTION

Microphthalmos is a developmental disorder of the eye due to arrest in growth of the ocular tissues. It is defined as a total axial length (TAL) at least two standard deviations below the normal TAL for the patient's age. Microphthalmos can be simple (without major ophthalmic complications) or mixed (presence of serious ophthalmic complications). Microphthalmos may primarily affect the posterior segment in which case it is termed as "posterior microphthalmos".[1] In previous reports, evaluation of anterior and posterior segment features of posterior microphthalmic eyes performed with A-scan and B-scan ultrasonography (USG),[2] electroretinogram (ERG), fluorescein angiography (FA),[1,2] Goldmann kinetic perimetry[3] and posterior segment optical coherence tomography (OCT) and ultrasound biomicroscopy (UBM).[4] With the advances in anterior segment (AS) imaging using OCT which provides high-resolution cross-sectional images, it is possible to explore AS structures more extensively.[5] To the best of our knowledge, no similar study reporting AS-OCT findings of posterior microphthalmos has been published. In this report, we aim to describe all the findings of posterior microphthalmos including AS-OCT.

CASE REPORT

Here we describe a case of a 21-year-old male patient who was admitted to our clinic with a history of low vision. There was no family history of eye disorders or he was not the product of a consanguineous marriage. The patient was otherwise healthy. On ocular examination, deep-set eyeballs were observed. Slitlamp examination showed slightly shallow anterior chambers with no additional pathologic findings. Dilated fundus examination revealed slightly elevated papillae with blurred disc margins (crowded optic discs), elevated horizontal papillomacular retinal folds [Figure 1a] and retinal pigment epithelium (RPE)

[1]Retina Clinic, Beyoglu Eye Research and Education Hospital, [2]Istanbul Medeniyet University, Istanbul, Turkey

Corresponding Author: Dr. Zeynep Alkin, Beyoglu Eye Research and Education Hospital, Bereketzade Cami Sok. No: 2, Beyoglu, Istanbul, Turkey. E-mail: zeynepalkin@gmail.com

migration in the mid-periphery of both fundi [Figure 1a-i]. On FA, markedly reduced diameter of capillary-free zone was noted [Figure 1b]. Furthermore, blocked fluorescence by pigment clumps and window defects in midperipheral fundus were observed on fundus fluorescein angiograms. Fundus autofluorescence imaging at 488 nm identified hypofluorescent linear horizontal markings corresponding to areas of papillomacular folds [Figure 1b-i]. Spectral domain OCT analysis of the macular region revealed that the papillomacular folds included neural retina without involvement of the RPE-choroid band [Figure 2a]. AS-OCT analysis revealed bilateral occludable narrow angles and thickened ciliary bodies [Figure 2b]. Full-field ERG testing showed generalized rod cone dysfunction associated with more prominent decrease in scotopic rod responses. B-mode USG demonstrated diffuse thickening of the choroid and sclera with foreshortening of the globe [Figure 3a]. Visual field was examined with the threshold 30-2 program of the Humphrey Field Analyzer (Carl Zeiss Meditec Gmbh, Jena, Germany) showing diffuse sensitivity loss and constricted peripheral visual fields [Figure 3b]. Optic nerve parameters with the Stratus OCT and lens thickness, TAL with A-scan USG are illustrated in Table 1. Additional AS parameters are presented in Table 1 including anterior chamber depth (ACD) and white-to-white corneal diameter by Zeiss IOL Master (Carl Zeiss Meditec Gmbh, Jena, Germany), corneal thickness and keratometric analysis by Orbscan II (Bausch and Lomb Inc., Rochester, NY, USA) and corneal endothelial cell count (ECC) with specular microscopy. As the main pathological features involved the posterior segment a diagnosis of posterior microphthalmos was established.

DISCUSSION

A wide variety of pathologic features can be associated with posterior microphthalmos. These features include pigmentary retinopathy, foveoschisis,[3] peripheral retinoschisis, retinal dialysis,[1] absence or reduction of capillary-free zone,[2] optic nerve head drusen,[3] papillomacular and/or chorioretinal folds,[2,3] and

Table 1: Additional anterior and posterior segment findings of the patient

	Right eye	Left eye
Refractive error (D)	+14.75-1.00 α65	+15.50-1.50 α130
BCVA	20/100	20/100
IOP (mm Hg)	12	10
Total AL (mm)	15.93	15.87
ACD (mm)	2.53	2.32
Lens thickness (mm)	4.43	4.38
WTWD (mm)	12.4	11.3
Keratometry (D)	K1=50.31 α65 K2=51.16 α155	K1=49.91 α130 K2=51.31 α40
CCT (μm)	584	634
ECC (cell/mm^2)	2351	2467
ACA (temporal/nasal)	5.5°/2.9°	2.4°/1.7°
Optic disc area	4426 mm^2	4661 mm^2
RNFL thickness	162 μm	155 μm

BCVA: Best corrected visual acuity, IOP: Intraocular pressure, AL: Axial length, ACD: Anterior chamber depth, WTWD: White to white distance, CCT: Central corneal thickness, ECC: Endothelial cell count, ACA: Anterior chamber angle, RNFL: Retinal nerve fiber layer

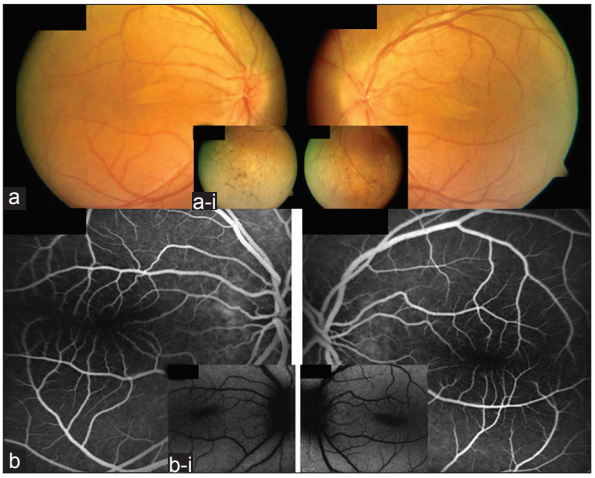

Figure 1: (a) Fundus pictures showing the crowded optic discs and elevated horizontal papillomacular retinal folds of the right and left eye. (a-i) Fundus view showing midperipheral retinal pigmentary changes in both eyes. (b) Fundus fluorescein angiographs showing the reduced capillary free zone of the right and left eye. (b-i) Blocked choroidal fluorescence by papillomacular folds in fundus autofluorescence imaging of both eyes

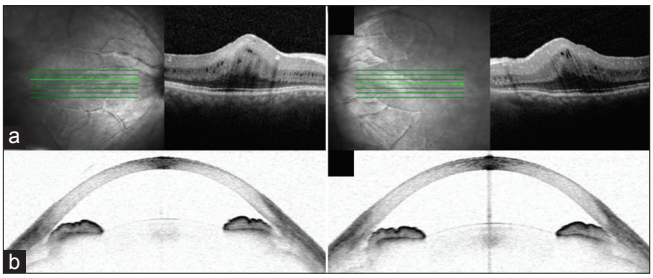

Figure 2: (a) Cross-sectional image on spectral-domain optical coherence tomography (OCT) of a patient revealing neural retinal folds of the right and left eye. (b) Narrow anterior chamber angles with thickened ciliary bodies in anterior segment-OCT imaging of the right and left eye

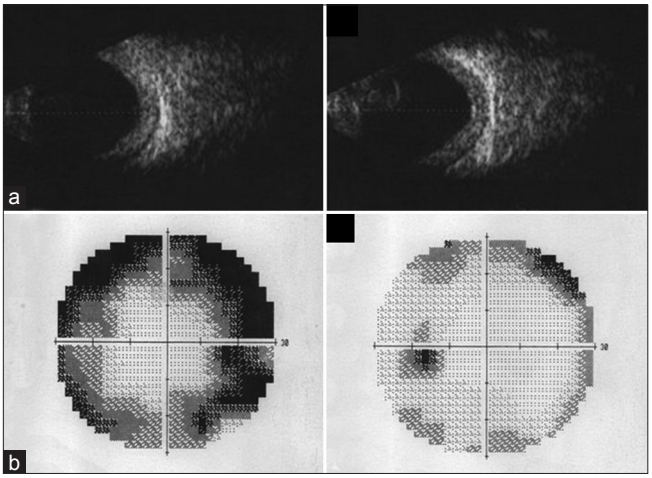

Figure 3: (a) Vitreous cavity of decreased length and sclerochoroidal thickening in B-scan ultrasonography for the right and left eye. (b) Representation of the constricted visual fields of the right and left eyes

uveal effusion syndrome.[6] In previous reports, some familial cases of posterior microphthalmos have been identified with autosomal recessive inheritance.[4] Based on negative family history and lack of consanguineous marriage, we believe that this is a sporadic type of microphthalmos. In prenatal eye growth, while the outer ocular coats including RPE, choroid and sclera lag behind in development, unrestrained neuroretinal growth causes papillomacular folds.[7] In our patient, OCT clearly demonstrated that papillomacular folds contained only neural retina without involvement of RPE and choroid as previously reported.[4] Although high hyperopia and elevated papillomacular folds are the main causes of visual impairment in posterior microphthalmic eyes, other chorioretinal changes such as pigmentary retinopathy should be considered as a contributing factor for limited vision. The present patient had RPE clumping suggestive of photoreceptor death which was confirmed by the ERG findings. Midperipheral scotomas are a characteristic feature of pigmentary retinopathies due to the loss of rod and cone function.[6] Visual field constriction and generalized depression in perimetric examination of our patient can be the contribution of either pigmentary retinopathy or high hypermetropia. We observed the presence of crowded optic discs in this patient, consistent with previous reports.[2] Optic nerve head analysis revealed increased retinal nerve fiber layer thickness and decreased optic disc area compared with patients with normal axial length. It is probably related to a dense arrangement of the nerve fibers in optic discs in the small scleral canal. Orbscan analysis showed the presence of steep corneas with high keratorefractive indices and increased corneal thickness. Corneal ECCs and lens thickness were similar to healthy age-matched subjects. Although ACD and horizontal corneal diameters were slightly lower in both eyes than in normal patients of this age, AS-OCT revealed nearly closed narrow anterior chamber angles (ACA), which can be a risk for acute angle closure. Erdol *et al*.[4] described similar anterior chamber characteristics in two posterior microphthalmic eyes with UBM. They[4] found displacement in ciliary bodies toward the pupil from their normal anatomic position and a subtle degree of supracohoroidal effusion. However in our patient, there was no evidence of pathologic changes in these structures except thickening in ciliary bodies and prominent narrowing of the ACAs. In this report, we described the complex ocular phenotype of posterior microphthalmos including pigmentary retinopathy, papillomacular folds, reduced capillary-free zone, thickened sclera and choroid as well as narrow ACAs. We found that not only posterior segment structures but also AS especially ACAs show abnormalities, which can be evaluated by AS-OCT. Clinicians should bear in mind that even if the microphthalmic features affect predominantly the posterior segment, anterior structures can be involved to a larger extentz than expected from the clinical examination. A complete ophthalmic examination and appropriate ancillary tests are necessary for the diagnosis of this complex disease and the recognition of all the possible ophthalmic complications.

REFERENCES

1. Kim JW, Boes DA, Kinyoun JL. Optical coherence tomography of bilateral posterior microphthalmos with papillomacular fold and novel features of retinoschisis and dialysis. Am J Ophthalmol 2004;138:480-1.
2. Khairallah M, Messaoud R, Zaouali S, Ben Yahia S, Ladjimi A, Jenzri S. Posterior segment changes associated with posterior microphthalmos. Ophthalmology 2002;109:569-74.
3. Ayala-Ramirez R, Graue-Wiechers F, Robredo V, Amato-Almanza M, Horta-Diez I, Zenteno JC. A new autosomal recessive syndrome consisting of posterior microphthalmos, retinitis pigmentosa, foveoschisis, and optic disc drusen is caused by a MFRP gene mutation. Mol Vis 2006;12:1483-9.
4. Erdol H, Kola M, Turk A, Akyol N. Ultrasound biomicroscopy and OCT findings in posterior microphthalmos. Eur J Ophthalmol 2008;18:479-82.
5. Jancevski M, Foster CS. Anterior segment optical coherence tomography. Semin Ophthalmol 2010;25:317-23.
6. Al Turki R, Baabbad R, Al Amro S. Posterior microphthalmos with uveal effusion managed by lamellar sclerotomies. Ann Ophthalmol (Skokie) 2008;40:193-6.
7. Boynton JR, Purnell EW. Bilateral microphthalmos without microcornea associated with unusual papillomacular retinal folds and high hyperopia. Am J Ophthalmol 1975;79:820-6.

Cite this article as: Alkin Z, Ozkaya A, Karakucuk Y, Demirok A. Detailed ophthalmologic evaluation of posterior microphthalmos. Middle East Afr J Ophthalmol 2014;21:186-8.

Source of Support: Nil, **Conflict of Interest:** None declared.