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Case report

Posterior microphthalmos with good visual acuity: A case report

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ABSTRACT

Purpose: We report the case of an 11-year-old boy with posterior microphthalmos who exhibited normal and age appropriate development of visual acuity.

Observations: At the initial diagnosis, when he was 3 years old, the best-corrected visual acuity (BCVA) was 20/125 in the right eye (OD) and 20/200 in the left eye (OS) with high hyperopia (cycloplegic refraction +15.75 D sphere OD and +16.25 D sphere OS). Eight years after he began wearing hyperopic glasses, BCVA was 20/16 OD and 20/20 OS. Optical coherence tomography did not reveal a foveal pit in either eye throughout the observation period. However, elongation of the outer segment and widening of the outer nuclear layers were observed.

Conclusion and Importance: Many cases of posterior microphthalmos demonstrate subnormal BCVA due to an abnormal foveal structure (papillomacular retinal folds, absence of the foveal pit and avascular zone) and high hyperopia. However, if foveal maturity progresses, even if the foveal structure is abnormal, early aggressive amblyopia treatment can result in normal and age appropriate development of visual acuity.

1. Introduction

Posterior microphthalmos (PM) is a rare type of microphthalmos that disproportionately affects the posterior ocular segment with a normal appearing anterior ocular segment.[1] Several cases of PM have been reported.[1–4] The principle findings of PM are high hyperopia and papillomacular retinal fold. In recent reports, biometric and molecular PM investigations have been performed, and the characteristics of PM have been described.[5,6] In these previous reports, PM was associated with subnormal visual acuity, and best-corrected visual acuity (BCVA) ranged from 20/25–20/200[1–6]. We examined a child with PM who exhibited normal and age appropriate visual development after eight years after the diagnosis and the case is discussed.

2. Case report

A 3-year-old boy presented to Toyama University Hospital (Toyama, Japan) with decreased vision in both eyes. There was no family history of ocular disease and his medical history was unremarkable. His physical and intellectual development were normal. BCVA was 20/125 in the right eye (OD) and 20/200 in the left eye (OS). Examination revealed high hyperopia (cycloplegic refraction +15.75 D sphere OD and +16.25 D sphere OS) and reduced total axial length (15.26 mm OD and 15.01 mm OS) as measured using an optical biometer. The patient had normal alignment, motility, and convergence testing. The eyes were deeply set but otherwise normal. Biomicroscopy findings from the anterior segment findings were unremarkable. The corneal powers were 50.8 D OD and 51.7 D OS. The central corneal thickness was 553 μm OD and 551 μm OS. The anterior chamber depth was 2.56 mm OD and 2.52 mm OS. The horizontal corneal diameter was 11.0 mm in both eyes. Posterior segment examination revealed bilateral elevated horizontal papillomacular retinal folds (Fig. 1). In macular spectral-domain optical coherence tomography (SD-OCT), there was no foveal pit in either eye (Fig. 2). Unlike normal fovea, all layers of the retina were present in the fovea. The folding included all layers of the neurosensory retina, except for the external limiting membrane and the photoreceptor layer. The patient was diagnosed with posterior microphthalmos and ametropic amblyopia. He was prescribed full cycloplegic hyperopic glasses to be worn full time. He was regularly examined in our hospital. His BCVA had developed gradually and reached to 20/20 in both eyes at age nine.

At age 11, 8 years after the first visit, BCVA was 20/16 OD and 20/20 OS. The intra-ocular pressure was 16 mmHg OD and 15 mmHg OS. The eye position was orthophoria. Axial lengths measured using an optical biometer were 15.23 mm OD and 15.02 mm OS, which were stable since the first visit. The anterior chamber depth was 3.06 mm OD and 2.96 mm OS. The horizontal corneal diameter was 11.46 mm OD

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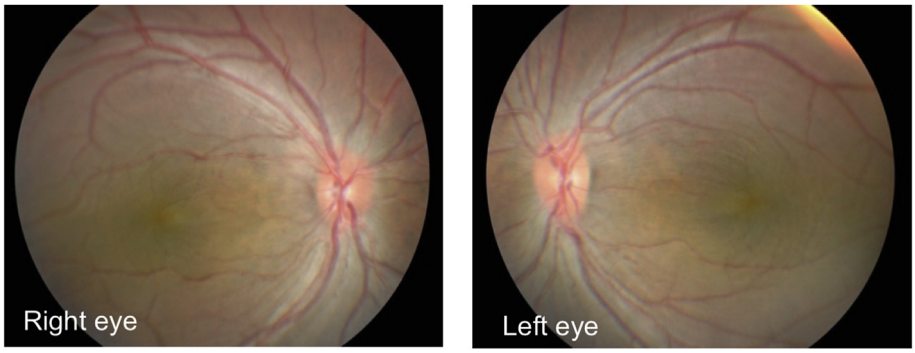

Fig. 1. Color fundus photograph of both eyes showing the papillomacular retinal fold when the patient was 3-year-old.

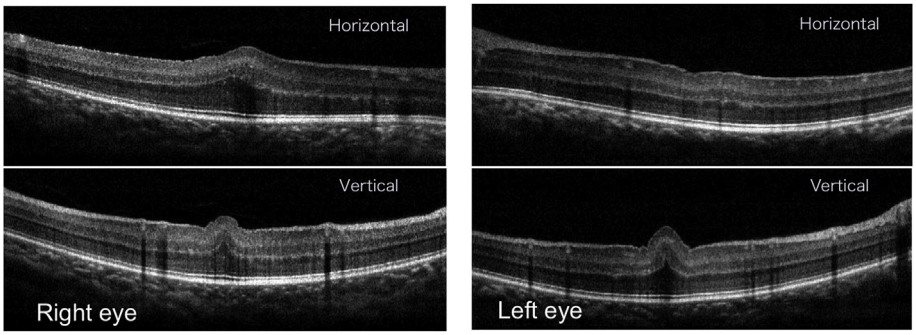

Fig. 2. In spectral domain optical coherence tomography image of both eyes showed the absence of foveal pit when the patient was 3-year-old.

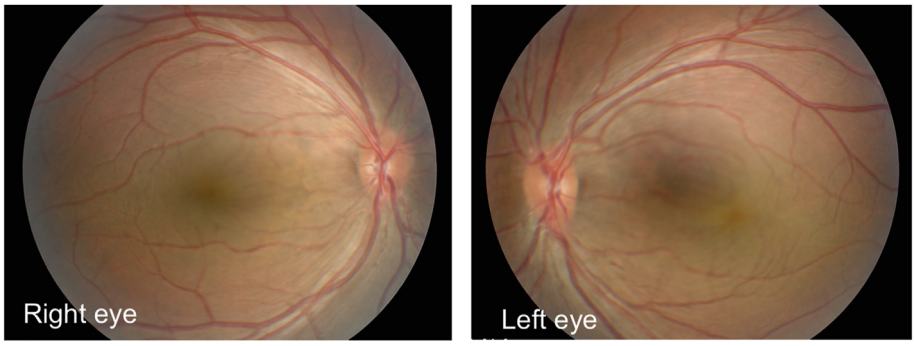

Fig. 3. Color fundus photograph of both eyes showing the papillomacular retinal fold when the patient was 11-year-old.

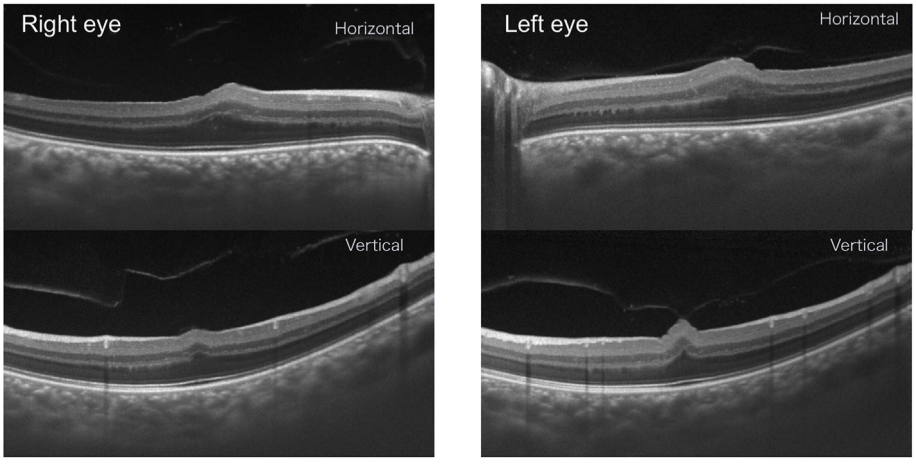

Fig. 4. In spectral domain optical coherence tomography image of both eyes showed the absence of foveal pit when the patient was 11-year-old.

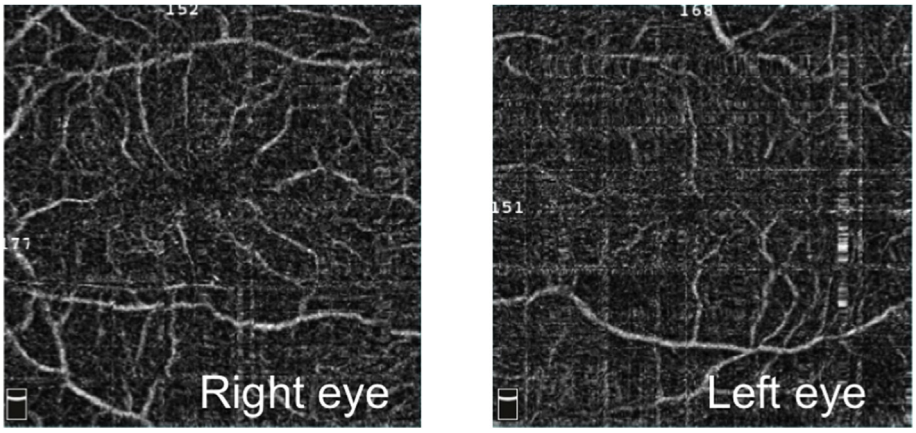

Fig. 5. Optical coherence tomography angiography of the fovea in the patient at 11-year-old. The corresponding B scan of each image shows segmentation at the level of superficial capillary plexus.

and 11.36 mm OS. The central corneal thickness was 566 μm OD and 569 μm OS. The corneal power was 50.9 D OD and 51.3 D OS. The lens thickness was 4.2 mm OD and 4.36 mm OS. There was no change in ophthalmoscopic foveal findings and SD-OCT compared to these at initial diagnosis (Figs. 3 and 4). There was no foveal pit in either eye throughout the observation period. However, elongation of the outer segment and widening of the outer nuclear layers (ONLs) were observed. Optical coherence tomography angiography (OCTA) did not clearly reveal a foveal avascular zone (FAZ) (Fig. 5). The patient could not be examined using multifocal electroretinography (mfERG) because his narrow conjunctival sacs precluded the patient from wearing contact lenses during mfERG.

3. Discussion

The principle findings of PM include high hyperopia and papillomacular retinal folds. In recent reports, biometric investigations of PM have been characterized and the details have been described.[5,6] The biometric findings and data from the present case were highly consistent with these reports. In previous studies, PM was associated with poor visual acuity, with BCVA ranging from 20/200 to $20/25^{1-6}$. Furthermore, in most PM patients with no complications, vision development failed even with continued refractive correction during childhood.[5] Vision impairment in PM is caused by high refractive amblyopia, absence of the foveal pit, papillomacular retinal folds, and other complications (uveal effusion, pigmentary retinopathy, chorioretinal folds).[1,5] However, the remarkable finding in our case was the normal and age appropriate development of BCVA. There is little doubt that diagnosis at an early age and appropriate amblyopia treatment in this case facilitated the development of normal and age appropriate visual acuity. A previously reported case of PM achieved normal and age appropriate visual development due to amblyopia treatment from an early age.[4] This emphasizes that poor visual development is not necessarily due to the lack of a foveal pit, but to foveal immaturity, especially the outer retina and refractive amblyopia.

OCT bands typical of adult fovea photoreceptors are absent near birth due to immaturity of the foveal cones but develop by 24 months and mature during childhood.[7] The foveal structure in the present case was comparable with the structural grading of foveal hypoplasia described by Thomas et al..[8] This grading was based on the presence or absence of a foveal pit, widening of the ONLs, and extrusion of plexiform layers and elongation of the outer segment. According to this grading, the present case would be classified as grade 2 given the absence of extrusion of plexiform layers, absence of a foveal pit, presence of outer segment lengthening, and presence of ONL widening. Thomas et al.[8] suggested that structural grading helps to provide a prognostic indicator for visual acuity. Grade 1 represents extremely good vision, and visual outcomes will become poorer as the grade increases.

On the other hand, Provis et al.[9] and Marmors et al.[10] reported that an anatomical foveal recess is not necessarily required for better visual acuity (BCVA ranged from 20/20 − 20/50 in the study by Marmors et al.[10]) and the reaction found in cone cell density at the fovea in adaptive optics (AO) and mfERG is important. In this case, however, cone cell density in the patient could not be counted with AO poor image due to his high hyperopia and poor fixation. In addition, the patient could not be examined using mfERG because he could not wear contact lenses of during the procedure due to his narrow conjunctival sac. Previous studies of FAZ characteristics in patients with nanophthalmos, using OCTA, reported that nanophthalmic FAZ was an unmeasurably small size.[11,12] In our patient, FAZ in both eyes was not revealed using OCTA as in previous studies.

Due to the presence of a relatively mature outer retinal structure and function, even without a foveal pit, it was suggested that vision in this case was likely to have developed due to photorefractive treatment from an early age. The patient described in this report is still a child; therefore, he will be carefully observed to determine if his current vision can be maintained. Appropriate refractive correction and diagnosis from an early age are important despite the presence of PM. In addition, a detailed observation of the foveal structure and function are helpful in predicting visual acuity prognosis of patients with PM.

Patient consent

The patient's legal guardian orally consented to publication of the case. This report does not contain any personal information that could lead to the identification of the patient.

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Authorship

All authors arrest that they meet the current ICMJE criteria for authorship.

Declaration of competing interest

All authors have no financial disclosures.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.ajoc.2019.100568.

References

1. Spitznas M, Gerke E, Bateman B. Hereditary posterior microphthalmos with papillomacular fold and high hyperopia. *Arch Ophthalmol.* 1983;101:413–417.
2. 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;138:480–481.
3. Aras C, Ozdamar A, Ustundag C, Ozkan S. Optical coherence tomographic features of papillomaacular fold in posterior microphthalmos. *Retina.* 2005;25:665–667.
4. Park SH, Ahn YJ, Shin SY, Lee YC. Clinical features of posterior microphthalmos associated with papillomacular fold and high hyperopia. *Clin Exp Optom.* 2016;99:590–593.
5. Khairallah M, Messaoud R, Zaouali S, Yahia SB, Ladjimi A, Jenzri S. Posterior segment changes associated with posterior microphthalmos. *Ophthalmology.* 2002;109:569–574.
6. Nowilaty SR, Khan AO, Aldahmesh MA, Tabbara KF, Al-Amri A, Alkuraya FS. Biometric and molecular characterization of clinically diagnosed posterior microphthalmos. *Am J Ophthalmol.* 2013;155:361–372.
7. Vajzovic L, Hendrickson AE, O'Connell RV, et al. Maturation of the human fovea: correlation of Spectral-domain optical coherence tomography findings with histology. *Am J Ophthalmol.* 2012;154:779–789.
8. Thomas MG, Kumar A, Mohammad S, et al. Structural grading of foveal hypoplasia using Spectral-domain optical coherence tomography. *Ophthalmology.* 2011;118:1653–1660.
9. Marmor MF, Choi SS, Zawadzki RJ, Werner JS. Visual insignificance of the foveal pit. *Arch Ophthalmol.* 2008;126:907–913.
10. Provis JM, Dubis AM, Maddess T, Carroll J. Adaptation of the central retina for high acuity vision: cone, the fovea and the avascular zone. *Prog Retin Eye Res.* 2013;35:63–81.
11. Funakoshi S, Yoshikawa T, Harada Y, Chikama T, Kiuchi Y. Absence of the foveal avascular zone in a nanophthalmic child revealed by optical coherence tomography angiography. *Am J Ophthalmol Case Rep.* 2019;13:34–37.
12. Mansour AM, Stewart MW, Yassine SW, et al. Unmeasurable small size superficial and deep foveal avascular zone in nanophthalmos: the Collaborative Nanophthalmos OCTA Study. Br J Ophthalmol. https://doi.org/10.1136/bjophthalmol-2018-312781.