# Posterior Microphthalmos with Papillomacular Retinal Folds and Pseudo Disc Edema

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## ABSTRACT

We report a rare case of posterior microphthalmos with prominent papillomacular folds, pseudo disc edema and high hyperopia in a 12 year old child with no systemic anomalies. The patient was diagnosed as high hyperopia two years ago but the posterior findings were missed. The anterior segment of the eye was normal on slit lamp examination but the appropriate diagnosis was missed. Hence detailed evaluation of every hyperopic child should be done for the timely management. This case report highlights the importance of detailed fundus and biometric evaluation to avoid missing the diagnosis of posterior microphthalmos in a high hyperopic child with no systemic anomalies.

## KEY WORDS

*Hyperopia, Microphthalmos, Papillomacular folds, Pseudo disc edema*

## INTRODUCTION

Posterior microphthalmos is a genetically heterogeneous congenital ocular malformation syndrome that comprises of papillomacular retinal fold and high hyperopia.[1] Posterior micophthalmos falls under the group of partial microphthalmos with normal anterior segment of eye.[2]

Posterior microphthalmos is associated with reduced bilateral axial length (15.55 – 18.61 mm), foreshortening of the posterior segment and a relatively normal anterior segment, high hyperopia (+11.0 to +15.75 Dioptre), retinal papillomacular folds, various types of esotropia, optic disc hypoplasia and neurosensory retinal detachment.[3]

We report a rare case of posterior microphthalmos with prominent papillomacular fold, pseudo disc edema and high hyperopia in a 12 year old child with no systemic anomalies. This case report highlights that the posterior findings might be missed owing to normal anterior segment of eye and absence of systemic anomalies in a case of high hyperopia without careful and detailed evaluation.

## CASE REPORT

A 12 years old boy came with chief complain of diminution of vision in both the eyes since childhood. The patient had no history of redness, eye ache, headache and photophobia in the past. There was no history of wearing glasses in the past.

The best corrected visual acuity was 3/60 in botheyes at the time of presentation. The retinoscopy value after cyclo-refraction was + 11.00 / -2.50 170° in the right eye and +11.00 / -2.00 180° in the left eye. The near vision

was N/32 in both teyes. The accepted glass power was + 6.00 Dioptre spherical lenses and the best corrected visual acuity with + 6.00 Dioptre (D) spherical lenses was 6/60 in both eyes. The intraocular pressure was eight mmHg in the right eye and 10 mmHg in the left eye at the time of presentation. The anterior segment examination was normal. The patient was advised for biometry and fundus examination but his parentsenied and insisted for glasses only. Provisional diagnosis of both eye high compound hyperopic astigmatism was made and glass was described.

The patient came every two yearly for follow up with complains of blurring of vision and glasses were continued. The retinoscopy value was same on every follow up and the patient had no systemic anomalies. After two years, the patient was sent for detailed fundus evaluation to retina clinic. The patient was sent for biometry and planned for dilated fundus examination. Dilated fundus examination showed decreased depth of vitreous chamber and presence of striae in the supero-temporal and infero-temporal area of the macula. The optic disc was crowded in nature and the disc margins were blurred. The optical coherence tomography of the optic disc and macula were normal. The papillomacular striae along with pseudo-disc edema were confirmed on fundus photo (Fig. 1) and optical coherence tomography. The papillomacular striae and pseudo disc edema led us to suspect posterior microphthalmos.

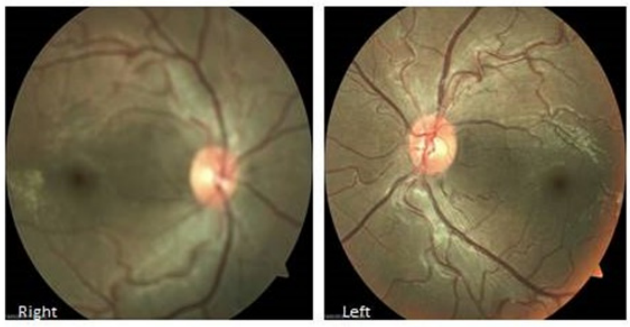


**Figure 1. Fundus photography showing papillomacular folds and blurred disc margins**

The patient underwent axial length measurement, keratometry, corneal pachymetry and detailed biometry evaluation as shown in table 1.

**Table 1. Biometry reading**

| Biometry | Value in right eye | Value in left eye |
|---|---|---|
| Axial length | 19.61 mm | 19.61 mm |
| Anterior chamber (ACD) | 3.21 mm | 3.23 mm |
| Lens thickness (LT) | 3.78 mm | 3.74 mm |
| Vitreous depth (VD) | 12.21 mm | 12.66 mm |
| Central corneal thickness (CCT) | 5.74 mm | 5.77 mm |
| Keratometry-reading | 40.96D/44.29D 170° | 40.91D/43.44D 0° |

The biometry showed normal anterior chamber depth (ACD), normal Lens Thickness (LT) but decreased Vitreous Chamber depth (VCD).

The positive findings of high hyperopia > 9.00 Dioptre, normal anterior chamber depth (ACD), normal lens thickness (LT), shorter axial length, decreased vitreous chamber depth (VCD), papillomacular striae and pseudpapilledema in the fundus of both eyes confirmed the diagnosis of posterior microphthalmos. The patient was prescribed same glasses and counselling was done for patching and no further intervention was required.

## DISCUSSION

Posterior microphthalmos is classified under partial microphthalmos with normal anterior chamber, normal cornea but small posterior segment in high hyperemetropic eyes.[2] Park et al. in his case series concluded that posterior microphthalmos was associated with multiple types of esotropia, optic disc hypoplasia, pappilomacular folds and neurosensory retinal detachment.[3] Early and appropriate management of the high refractive error and esotropia is essential particularly in children with posterior microphthalmos.[3] This is a case of 12 year old boy whose diagnosis was missed as refractive error with high compound hyperopic astigmatism for two years. He was finally diagnosed as posterior microphthalmos with papillomacular folds only after detailed retina evaluation. The retinoscopy value after cyclo-refraction was + 11.00 D / -2.50 Dcy 170 degree in the right eye and +11.00 Dsp / -2.00 Dcy at 180 degree in the left eye and the best corrected visual acuity was 6/60.

Park Shin reported four cases with high hyperopia with +11.0 to +15.5 Diopter similar to our case. There was also reduced bilateral axial length and foreshortening of the posterior segment but relatively normal anterior segment.[2] The anterior chamber depth in our case was also normal with value of 3.21 mm in right eye and 3.33 mm in left eye. The lens thickness was also normal in our patient. Our patient had reduced axial length of 19.61 mm in right eye and 19.69 mm in left eye. Keratometry showed 40.96D / 44.29 D at 170 degree in right eye and 40.91D / 43.44D at 0 degree in the left eye. The Central corneal thickness was 5.74 mm in right eye and 5.77 mm in left eye. The vitreous depth was reduced with a value of 12.21 mm in right eye and 12.66 mm in left eye.

Nowality et al. also found the mean cycloplegic refraction of +15.09D (Range 9.88 - 18.75D) similar to ours.[4] They also described that papillomacular fold in the retina may be due to excess tissue formed during normal growth of retina and under growth of sclera.[4] Khairallah et al. in his study had similar features to our patient with bilateral foreshortening of the posterior ocular segment (range, 7 – 11.2 mm) with associated high hyperopia (range, 12.00 – 19.00 diopters) and normal or slightly smaller than normal anterior segment dimensions.[7]

Posterior segment changes in posterior microphthalmos are bilateral elevated papillomacular retinal fold; fine retinal folds; chorioretinal folds; uveal effusion syndrome; pigmentary retinopathy, including retinitis punctata albescens, crowded optic discs; and sclerochoroidal thickening on ultrasonography. The posterior microphthalmos, retinitis pigmentosa and optic disc drusen syndrome along with whitish spots in posterior pole has been described. Systemic anomalies like cleft lip may be present unlike our case.[5-7] The posterior segment in our patient had prominent bilateral papillomacular retinal folds and pseudo disc edema. Zor et al. also detected bilateral retinal folds in six patients similar to ours but one of their cases had pigmentary retinopathy.[8] We did not detect any pigmentary retinopathy unlike Ramirez et al. case of posterior microphthalmos with fundus finding of retinitis pigmentosa, foveoschisis and optic disc drusen.[9]

This case report highlights the importance of detailed fundus and biometric evaluation to avoid missing the diagnosis of posterior microphthalmos in a high hyperopic child.

Posterior microphthalmos with prominent papillomacular retinal folds and pseudo disc edema may be missed in children with high hyperopia without detailed fundus evaluation and biometric tests due to normal anterior segment of eye and absence of systemic anomalies. Hence, detailed evaluation of every hyperopic child should be done for the timely management.

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