Case Report

Posterior Microphthalmia, A Challenging Diagnosis

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Microphthalmia involves eyes with total axial length of at least 2 standard deviations below age-similar controls. This case report presents an unusual form of microphthalmia, the posterior microphthalmia which has never been reported in Pakistan before. It also emphasises on the importance of use of Optical coherence tomography (OCT) for the diagnosis of posterior micropohthalmia. A 7 year old boy presented to us with bilaterally decreased vision and was found to have bilateral high hypermetropia. His fundal examination showed blurred optic disc margins and dolphin - shaped elevated pappillomacular fold extending from the fovea to the optic disc in both the eyes. OCT showed elevated neurosensory retina with normally attached retinal pigment epithelium. This confirmed the diagnosis of posterior Microphthalmia. The use of OCT thus aids in not just establishing the diagnosis of posterior microphthalmia but also prevents us from developing the wrong diagnosis of papilledema and carrying out any unnecessary investigations.

Key words: Posterior microphthalmia, High hypermetropia, Optical coherence tomography, Pseudopapilledema.

Microphthalmia is defined as total axial length of eyeball at least 2 standard deviations below age – similar controls. Posterior microphthalmia is a rare subset of microphthalmia in which the Total axial length of the eye ball is reduced resulting in high hypermetropia. Whilst the anterior segment dimensions including corneal diameter, anterior chamber depth and anteroposterior length of the lens are normal, the posterior segment is foreshortened and is associated with a papillomacular retinal fold. The optic discs are crowded with blurred margins.[1,2] This case report highlights the significance of Optical Coherence Tomography in diagnosing this rare entity. To our knowledge no case of Posterior Microphthalmia has been reported in Pakistan before.

CASE REPORT

A 7 year old boy presented to us in June 2014 with the complaint of gradually progressive loss of distant vision in both eyes for the last 2 years. He had been using spectacles for the last 2 years. His medical, surgical, drug and birth history were un-eventful. His parents had had consanguineous marriage.

On examination visual acuity was 1/60 unaided OU (both eyes) and it improved to 6/12 with +10.00 DS (diopter spherical) OD (right eye) and 6/18 OS (left eye) with +11.00 DS, respectively. His anterior segment examination was unremarkable with the corneal diameter within normal range, Keratometry-readings OD: K1: 47.25 × 90° and K2: 47.25 × 180° OS: K1: 47.00 × 90° and K2: 47.25 × 180°, Central Corneal Thickness OD 0.591 microns and OS 0.589 microns. The anterior chamber depth was 3.06 mm OD and 3.07 mm OS. Lens thickness was 3.50 mm OU**.** Axial length OD was 16.5 mm and OS 17.00 mm which were smaller for his age.

On fundus examination we found small clustered optic disc with blurred margin OU and a dolphin shaped elevated papillomacular retinal fold extending

from the fovea to the optic disc OU (Fig. 1). Intraocular pressure was 12 mm Hg OU. Optic nerve function tests were normal in both the eyes as were the macular function tests. Extra-ocular muscle movements were full in all gazes. Systemically the patient was normal and did not complain of headache or vomiting as might be expected with bilateral optic disc swelling when suspecting for Papilledema.

To study this unusual finding of the elevated papillomacular retinal fold, Ultrasound B-Scan and optical coherence tomography (OCT) were perfomed. The Ultrasound B-Scan revealed small eye balls with foreshortened posterior segment. OCT was done to further study the elevated retinal fold closely and it showed that the neurosensory retina was folded as a unit leaving the RPE and the choroid normally intact. There were cystic lesions present within this fold OU. (Fig. 1) There was no other associated pathology in the retina. The patient's siblings did not reveal similar ocular findings.

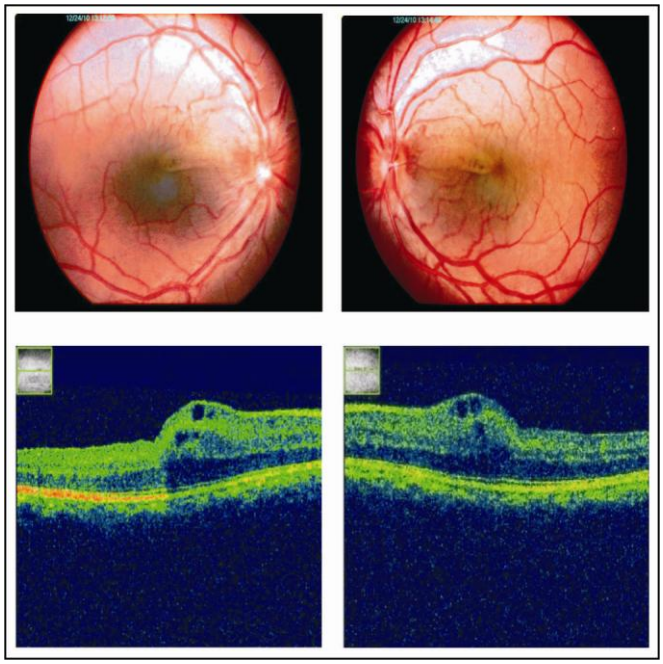

Fig. 1:
Top; Right: Fundus photo OD, Left: Fundus photo OS. Both showing blurred optic disc margins and dolphin shaped pappillomacular retinal fold between the optic disc and the fovea.
Bottom Right: OCT OD, Left: OCT OS. Both showing elevated neurosensory retina with contained cystoids spaces.

DISCUSSION

Posterior microphthalmia is a rare subset of microphthalmia in which the anterior segment of the eye is within normal dimensions but the posterior segment is foreshortened.[1,2] The total axial length is thereby reduced. This results in severe hypermetropia with significant associated clinical findings. The most prominent finding is bilaterally small optic discs with blurred margins and an elevated papillomacular retinal fold of the neurosensory retina whereas the RPE and choroid remain intact. This is attributed to the fact that the sclera is abnormally thickened; limiting the growth of RPE and the choroid, while allowing normal growth of the neurosensory retina.[3]

The blurred margins of the optic discs might give the false impression of papilledema but since intracranial pressure is normal in these patients thereby this is labeled under pseudopapilledema. Autosomal recessive and sporadic patterns have been reported for this syndrome.[1]

Many other clinical associations have been reported by various authors, along with these hallmark findings. These include; retinoschisis, dialysis,[4] esotropia, optic nerve hypoplasia[5], chorioretinal folds, uveal effusion syndrome, pigmentary retinopathy, retinitis punctata albescens, absent or marked reduction of the capillary - free zone[1], Duane retraction syndrome[7] and iridocorneal anomaly.[4-7]

High hyperopia and elevated papillomacular retinal fold are the main causes of visual impairment in such children.

Newer investigations like Ultrasound biomicroscopy (UBM) and optical coherence tomography (OCT) have not only helped in studying the nature of posterior microphthalmia but also the various anterior and posterior segment clinical findings found associated with it.

In our case we used OCT to study the papillomacular folds. It is an advancement of the recent past. It gives cross section of the retina and thereby clearly elicits each of its layers. Any retinal pathology can hence be extensively studied on OCT in aspects of its level, nature and type.

Posterior microphthalmia is a challenging diagnosis. Its timely diagnosis is critical to prevent the patient from the misdiagnosis of papilledema and thus unnecessary radiological investigations. OCT is a very helpful advancement which aids in seeing the cross-

sections of retina. In our case it clearly showed the elevated neurosensory retina and the flat retinal pigment epithelium thus helping us in reaching the diagnosis of posterior microphthalmia. Its use is strongly advocated in cases suspected for posterior microphthalmia.

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