Evaluation of Visual System in Microphthalmic Eyes

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Abstract

Background: Microphthalmia is defined as a globe with a total length that is at least two standard deviations below the mean for age. It can be isolated or occur with other anomalies or as part of a syndrome.

Objective: The aim of our work was to evaluate any functional or structural abnormality along the whole visual system in microphthalmic eyes.

Patient and Methods: The study included 70 eyes of 42 individuals having microphthalmos. The mean age was 12.47 years (ranged from 9 months to 15 years). All cases were subjected to full ophthalmological history, and examination. Some investigative tools were used as A scan ultrasound, ultrasound biomicroscopy (USB), optical coherence tomography (OCT), visually evoked potential (VEP), and impression cytology.

All data were recorded and analyzed.

Results: Positive family history was present in 29 cases (69%) while positive consanguinity was present in 24 cases (58%). Axial length ranged from 12.8 to 19.9mm (mean 16.1 ± 2.3). Simple microphthalmos (without any ocular dysgenesis) was recorded in 9 cases (21.4%). Nystagmus was present in 19 cases (45.2%), convergent squint in 9 cases (21.4%), Congenital contract was present in all eyes included in this study. Other ocular dysgenesis found were microcornea 28 eyes (40%), iris coloboma in 3 cases (7.2%), sclerocornea in 1 patient bilaterally (2.4%), posterior synechiae with pinpointed pupil in 11 eyes (15.7%), patches of iris atrophy in 3 cases (7.2%). Increased intraocular pressure with pallor optic nerve was present in 32 eyes (45.7%). Retinal examination showed picture like retinitis pigmentosa in 3 cases bilaterally (7.2%). Venous congestion and arterial attenuation with decrease vascular reflex were evident in 12 eyes (17.2%). VEP showed marked decreased in signals in all the cases which denotes functional defect of optic nerve.

UBM confirmed the presence of congenital cataract in all the cases (100%). Persistent hyperplastic primary vitreous (PHPV) was encountered in 3 cases (7.2%), corneal hyperreflectivity with wrinkling at Descemet's membrane in 1 case (2.4%), and peripheral anterior synchiae was present in 2 eyes (2.9%), while abnormal shaped angle was found in 3 eyes (4.3%).

Impression cytology showed marked squamous metaplasia (Grade IV) in almost all cases with completely absent goblet cells.

Conclusion: Microphthalmic eyes comprise large entity ranging between simple microphthalmos to complex type. Thus ophthalmological evaluations using examinations and different investigative tools help in suitable management together with control of the necessity of any surgical intervention and perform an accurate follow-up.

Key Words: Microphthalmia – Microcornea – Dysgenesis.

Introduction

MICROPHTHALMIA is defined as a globe with a total length that is at least two standard deviations below the mean for age. Classification of microphthalmia is according to the anatomic appearance of the globe and severity of axial length reduction.

Severe microphthalmia refers to a globe with a corneal diameter less than 4mm and a total length less than 10mm at birth or less than 12mm after the age of one year. Simple microphthalmia refers to an eye that is anatomically intact except for its short total axial length, while complex microphthalmia is that eye with anterior or posterior segment dysgenesis [1].

Microphthalmia can be unilateral or bilateral. It is a heterogeneous condition with various etiologies. It can be isolated or occur with other anomalies or as part of a syndrome [2].

Patients and Methods

The study was conducted in Ain-Shams University Hospital during the period from June 2008 to March 2010. The study included 70 eyes of 42 individuals having microphthalmic eyes. The mean age was 12.47 years (ranged from 9 months to 15 years).

The eye was considered to be microphthalmic if the axial length was less than 20mm. Patients
with normal axial length were excluded from the study. (Relative anterior microphthalmos was considered when the anterior segment was small with microcornea and normal axial length).

All eyes were subjected to full ophthalmological examination including measurement of the corneal diameter and anterior segment evaluation for the presence of associated eye anomalies. Measurement of intraocular pressure and fundus examination were done if possible.

Axial length was measured under general anesthesia using A scan ultrasound. USB was done to determine all anterior segment abnormalities if present. It was performed using Humphrey instrument with 50-MHZ transducer probe allowing 4-5mm tissue penetration.

OCT was done to study the posterior segment in microphthalmic eyes.

VEP was also done to assess optic nerve function.

Impression cytology was also done to assess any micro structural abnormalities in the ocular surface. All data were recorded and analysed.

Results

Seventy eyes of 42 children were included in this study. Family history was positive in 29 patients (69%) and positive consanguinity was found in 24 patients (58%). The age ranged from 9 months to 15 years (<1 year = 10 cases, 1-6 years = 21 cases, 6-10 years = 5 cases and 10-15 years = 6 cases).

Nystagmus was present in 19 cases (45.2%), convergent squint in 9 cases (21.4%) and divergent squint was not recorded in our study. Congenital contract was present in all eyes included in this study varies from dot-shaped cortical opacities 8 eyes (11.4%), anterior polar cataract 2 eyes (2.9%), posterior polar cataract 60 eyes (85%), and nuclear cataract 2 eyes (2.9%).

Other ocular dysgenesis found were microcornea 28 eyes (40%), iris coloboma in 3 cases (8.6%), sclerocornea in 1 patient (2.4%), posterior synechia with pin-pointed pupil in 11 eyes (15.7%), patches of iris atrophy in 3 cases (8.6%). Increased intraocular pressure with pallor optic nerve was present in 32 eyes (45.7%). Retinal examination showed picture like retinitis pigmentosa in 3 cases (8.6%). Venous congestion and arterial attenuation with decrease vascular reflex were evident in 12 eyes (17.2%).

A scan ultrasound showed that axial length ranged from 12.8 to 19.9mm (mean 16.1 ± 2.3) (Fig. 1). Simple microphthalmos (without any ocular dysgenesis) was recorded in 9 cases (21.4%) (Fig. 1).

UBM confirmed the presence of congenital cataract in all the cases included in this study which appears echographically as opaque swollen lenses. It varies from dot-shaped cortical opacities 8 eyes (11.4%), anterior polar cataract 2 eyes (2.9%), posterior polar cataract 60 eyes (85%), and nuclear cataract 2 eyes (2.9%).

Persistent hyperplastic primary vitreous (PHPV) was encountered in 3 cases (7.2%). Also UBM showed corneal hyper reflectivity with wrinkling at Descemet's membrane in 1 case (2.4%), and peripheral anterior synichea (PAS) was present in 2 eyes (2.9%), while abnormal shaped angle (angle dysgenesis) was found in 3 eyes (4.3%) (Fig. 2).

VEP showed marked decreased in signals in all the cases which denotes functional defect of optic nerve.

Impression cytology showed marked squamous metaplasia (Grade IV) in almost all cases with completely absent goblet cells in all cases.

OCT showed no abnormalities except in 9 cases (21.4%) with alternating retinal nerve fiber reflectivity being thin in one area and thick in another.

Fig. (1): A scan ultrasound showing decrease in axial length.
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Fig. (2): USB showing narrow angle and PAS.

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<th>Type of investigation</th>
<th>Number of cases</th>
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<td>19 cases</td>
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<td>USB findings</td>
<td>3 cases</td>
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<td>PAS</td>
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<td>Corneal hyperreflectivity</td>
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<td>Venous cong &amp; arterial attenuation</td>
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<td>17.2%</td>
<td>OCT</td>
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Discussion

Microphthalmos is a developmental disorder in which the eye is smaller than normal eyes. This disorder can present as an isolated condition or associated with other systemic alterations [3].

Microphthalmos probably represents a nonspecific growth failure in response to a very wide variety of prenatal insult which include, hereditary, chromosomal aberrations and unknown causes [4].

Seventy eyes of forty two children with microphthalmos were included in this study 69% showed positive family history while positive consanguinity was present in 58%.

Congenital cataract was present in all cases with variable site, shape and density, but the most encountered type was dense posterior polar cataract (83%). Elder [8] study revealed that the incidence of congenital cataract among microphthalmic eyes was 44% in his study. Yu, et al. [2] reported that cataract surgery must be done carefully to avoid complications as secondary membranes and corneal opacities.

Sturmer, et al. [6] claimed that the small anterior segment in microphthalmos especially the shallow anterior chamber and the presence of small undilated pupil are responsible for major intraoperative complications during cataract extraction in these patients.

Elevation of intraocular pressure with the presence of narrow angle with or without anomalies was encountered in 30 of our patients. In agreement with our study, Auffarth, et al. [7] reported that there was high incidence of narrow angle and angle closure glaucoma in these microphthalmic patients.

Posterior segment examination and echography showed pallor optic nerve in 30 cases, retinitis pigmentosa-like changes in 6 eyes, vascular changes in 12 cases and PHPV was echographically encountered in three cases bilaterally in our study. While kairallah, et al. [8] found that a subset of individuals can have visual loss resulting from posterior segment abnormalities including papillo-macular folds, macular hypoplasia, cystoid macular edema and uveal effusion.

The echographic features encountered in this study are not different from those widely reported
in other research. Previous studies had reported that microphthalmos may be associated with severe eye diseases as congenital cataract, PHPV and coloboma. Elder [5] study showed 40.74% incidence of PHPV in microphthalmos eyes while Weiss, et al. [9] diagnosed 30% of PHPV in their research.

In our study we didn’t find any case with choroidal coloboma, while Weiss, et al. [9] found 35% of his studied cases had choroidal coloboma.

UBM was found to be more effective in detecting peripheral anterior synaechia and iris atrophy than slit-lamp microscopy and gonioscopy, mainly because of corneal opacities in some anomalies associated with microphthalmos [10].

According to our knowledge this is the first time to do OCT, visual evoked potential and impression cytology in microphthalmic eyes which are valuable to indicate functional defect of optic nerve and any ocular surface abnormalities.

Conclusion:

Microphthalmic eyes comprise large entity ranging between simple microphthalmos to complex type. Thus ophthalmological evaluations using examinations and different investigative tools help in suitable management together with control of the necessity of any surgical intervention and perform an accurate follow-up.

References