

A Rare Case of Microphthalmos with Anophthalmos

ABSTRACT

Unilateral anophthalmia is an extremely rare congenital anomaly having prevalence of around 0.003%.^[1] Congenital microphthalmia is again a rare anomaly having prevalence rate of around 0.014%–0.035% and affects 3%–11% of blind children.^[1-3] The objective is to report a case of a 5-year-old male showing clinical right-sided microphthalmos and left-sided anophthalmos.

Key words: Anophthalmia, Child, Empty socket, Microphthalmos

INTRODUCTION

A severe developmental disorder of the eye wherein the eye is abnormally small and anatomically malformed is called microphthalmia or microphthalmos.^[1] Whereas anophthalmos is a condition where there is no evidence of ocular tissue in the orbit.^[1] Both these conditions could be unilateral or bilateral.^[1] Some of the common etiological factors responsible for both these conditions could be maternal infections during pregnancy, for example, toxoplasma, rubella, certain strains of influenza virus, varicella, cytomegalovirus, and parvovirus B19.^[4-11] Maternal Vitamin A deficiency, alcohol abuse, X-rays exposure, fever, hyperthermia, and drug intake during pregnancy like thalidomide are also found to be responsible for both these conditions. Mutations in genes SOX2, OXT2, and PAX6 are some of the gene defects and chromosomal anomalies like Trisomy 13 are common in both conditions. Triploid syndrome, Wolf–Hirschhorn syndrome, and 13q deletion are seen to be associated with microphthalmia.^[12] Whereas deletion of chromosome 14, defect in gene CHX10 and RAX, Lenz syndrome, Waardenburg syndrome are found to be associated with anophthalmia more frequently.^[1]

CASE REPORT

A 5-year-old child presented to the ophthalmology outpatient department with complaints of diminished vision in the right eye and no vision in the left eye since birth, also there were complaints of watering from the left eye. There was no history of fever during pregnancy of mother [Figure 1]. There was a Grade III consanguineous marriage, between the parents however, the older sibling, a girl child, was normal [Figure 2]. No medical or systemic illness was present. Parents gave history that the child was not able to open the eyes properly on his own and occasional redness of the right eye. On examination, the vision in the right eye was perception of light, projection of rays could not be assessed. On the left side,

Priyanka Dileep Asgaonkar

Department of Ophthalmology, Pravara Institute of Medical Sciences, Ahmednagar, Maharashtra, India

Corresponding Author:

Priyanka Dileep Asgaonkar,
Department of Ophthalmology, Pravara Institute of Medical Sciences,
Ahmednagar, Maharashtra, India.
E-mail: priyanka.asgaonkar@gmail.com

the eyeball was absent. The microphthalmic right eye showed horizontal nystagmus [Figure 3]. A small swelling about 1 cm × 1 cm in size was observed under the left lower lid, which on palpation was painless with the left socket being empty and no evidence of any ocular tissue, rest details were not appreciated. Clinically, there was no any other evidence of systemic associations such as craniofacial defects, microcephaly, mental retardation, seizures, webbing of hands or feet, external ear defects, and polydactyly. A magnetic resonance imaging (MRI) (brain and orbit) was performed which showed bilateral eye globes to be small in size with axial length being 11 mm (right eye) and 10 mm (left eye). A 2.5 cm × 1.9 cm × 2.3 cm multilobulated, intraorbital, extraconal cystic lesion was seen in the inferior half of displacing the left globe superiorly. Similar morphology lesion of 1.2 cm × 1.1 cm × 1.3 cm (AP × TRA × CC) was seen in intraconal compartment of the right orbit in retrobulbar location. Diffuse T2W and flair hyperintensities of periventricular and subcortical white matter, delayed myelination, and venous angioma in right parietal lobe of brain were also observed. Thus, the final impression of the MRI report concluded the case to be suggestive of the right eye microphthalmos and left eye anophthalmos.

Radiologically (as per the MRI reports), the axial length of the right eye was <20 mm which correlated with the clinical features of small size of eyeball, thus confirming the diagnosis



Figure 1: Right-sided microphthalmos



Figure 2: Left-sided anophthalmos



Figure 3: Both eyes together

of the right eye microphthalmos; also, the MRI finding of complete failure of outgrowth of the primary optic vesicle in the left eye correlated with the clinical features of the left eye globe absence, thus confirming the diagnosis of the left eye anophthalmos. Further management was not possible as there was loss to follow-up on the part of patient.

DISCUSSION

A complete regression of optic nerve vesicle causes a true anophthalmos, the condition being rare is diagnosed only on a serial histological sectioning of orbit on computed tomography (CT) scan, MRI, or ultrasound B scan, which would reveal a complete absence of ocular tissue in the orbital socket.

Clinically, anophthalmos could be primary, secondary, or a consecutive (degenerative) condition.^[13] when there is failure of optic vesicle development with no gross anomalies of the medullary tube, it is called primary anophthalmos.^[11] When there is complete suppression or gross abnormal development of entire part of the neural tube, the condition is called secondary anophthalmos, this condition is usually not compatible with life due to its association with severe malformations.^[11] In consecutive type of anophthalmos, the formed optic vesicle undergoes degeneration subsequently.^[11] When the size of the eye is below two standard deviation as compared to normal eye along with anatomical malformation, it is identified as microphthalmia.^[14] Depending on the presence of uveal coloboma, microphthalmia can be classified as colobomatous or non-colobomatous.^[14] Ocular findings seen in microphthalmia may be microcornea, opacity, ectopia lentis, aniridia, corectopia, persistent fetal vasculature, and retinal dysplasia or the disease may be associated with a cyst arising from optic nerve.^[14] When there is retinal or optic nerve involvement, the visual outcome is poorer as compared to when there is a presence of small iris or choroidal coloboma. Microphthalmia may be associated with mental retardation, polydactyly, or craniofacial malformations. Although most cases are found to be occurring sporadically some of them have been documented, some of them are documented to be inherited as autosomal dominant, autosomal recessive, or X linked.^[1] Ultrasonography, CT scan, and MRI are the mainstay investigations in anophthalmia and microphthalmia, additionally, VEP can be performed in microphthalmos to assess the visual function.

The management of patients should be composed of evaluation by multidisciplinary teams of pediatricians, ophthalmologists, and clinical geneticists as these conditions have a variable phenotypic spectrum. In microphthalmos when retinal function is detectable, refraction and treatment of amblyopia are of utmost value. Reconstructive strategies aim simultaneous management of both soft-tissue hypoplasia and asymmetric bone growth.^[15-17] Anophthalmic socket is treated with a prosthetic eye or with socket expanders.^[14]

CONCLUSION

With regard to case discussed above, it is significant that timely evaluation and early intervention in children with microphthalmos with minimal retinal function will have a huge impact on their visual prognosis. Furthermore, cosmetic management of anophthalmos will help the affected children to lead a normal life.

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