A Rare Case of Left Sided Anophthalmos with Congenital Cystic Eyeball with Right Sided Microphthalmos

Umesh Harakuni, Smitha K.S., Nagbhushan Chougule, Madhura Patil, Preetha Sudhakaran, Maria Sequeira Linda
Department of Ophthalmology, Jawaharlal Nehru Medical College, Belgaum, Karnataka, India

Summary

Congenital cystic eye, also known as anophthalmos with cyst, is an extremely rare congenital anomaly, first described by Mann in 1939 with a prevalence of 3 per 1,00,000. Congenital micropthalmos is also a rare condition with prevalence rates of 1.4 - 3.5 per 10,000 births. The objective is to report a case of a 19 year old male who was born to non-consanguineous marriage, presenting with left sided anophthalmos with right sided microphthalmos, with horizontal nystagmus and healed perforated corneal ulcer. Differential diagnosis include encephalocele, dermoid cyst, congenital cystic eye and tumors. Early visual rehabilitation is advised.

Delhi J Ophthalmol 2019;29;86-88; Doi http://dx.doi.org/10.7869/djo.430

Keywords: *Anophthalmos, Microphthalmos, Congenital cystic eyeball*

Case Report

A 19 year old male patient presented to ophthalmology OPD with the complaints of diminution of vision in both eyes since birth. There was history of fever during the first trimester of pregnancy for which the mother had taken some medication from a local doctor. There was no history of consanguineous marriage, other sibling was normal. No other chronic medical or systemic illness was present. Clinically, there was no evidence of systemic associations like facial defects, webbed hands, colobomatous lids, preauricular tags, hydrocephalus/ microcephaly, seizures or cleft lip. There was on and off history of pain and redness in the right eye. On examination, vision in the right eye was perception of light and accurate projection of rays and on the left side, the eye ball was absent. On examination, microphthalmos and horizontal nystagmus was present in the right eye with healed perforated corneal ulcer with iris incarceration. The remaining details were not made out. The left orbit was empty with remnants of conjunctiva and soft tissue. A 2 cm × 2 cm diffuse swelling was present near the left inferior orbital margin (Figure 1,2). On B scan ultrasonography, the right eye showed an axial length of 16.96 mm with some vitreous opacities and in the left eye, B scan showed the presence of a cystic lesion, with axial length 26.39 mm (Figure 3,4).

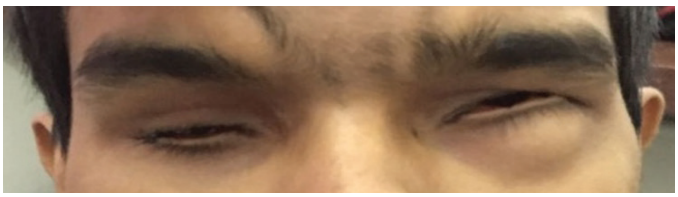

Figure 1

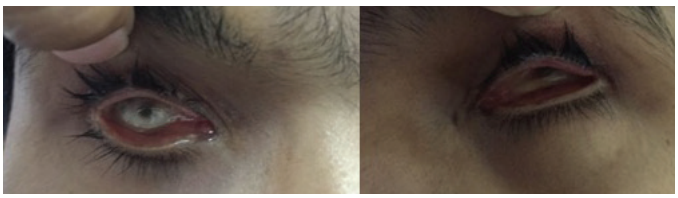

Figure 2: *(a) Right Eye showing microphthalmos with perforated corneal ulcer (b) Left Eye showing a 2cm × 2cm diffuse swelling near left inferior orbital margin.*

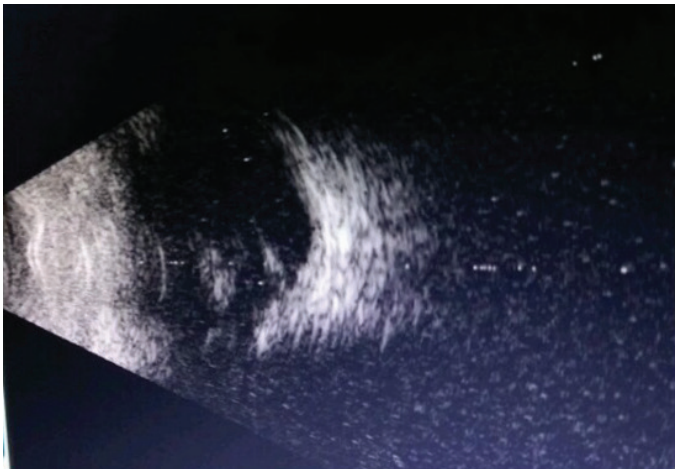

Figure 3: *Right eye B scan showing vitreous opacities with microphthalmos.*

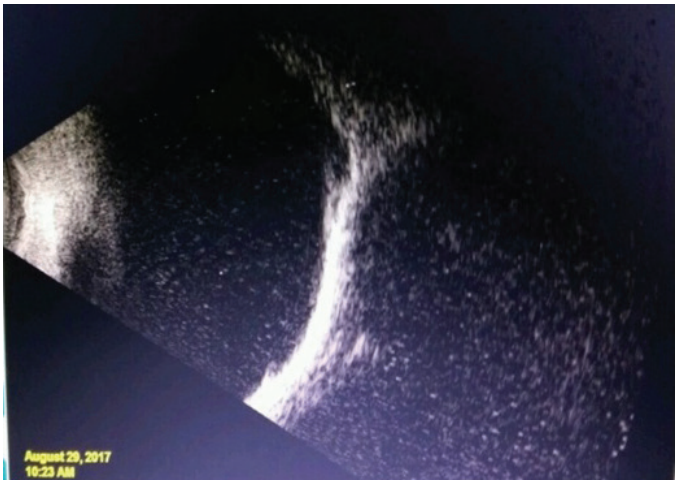

Figure 4: *Left eye B scan showing a cystic lesion.*

Discussion

Congenital cystic eye or anophthalmia with cyst is an unusual benign congenital malformation of the eye.

The International Clearinghouse for Birth Defects Monitoring Systems defines anophthalmia and microphthalmia as "anophthalmos/microphthalmos: apparently absent or small

eyes. Some normal adnexal elements and eyelids are usually present. In microphthalmia, the corneal diameter is less than 10 mm, and the antero-posterior diameter of the globe is less than 20 mm".[1]

Bardakjian et al, described anophthalmia as a complete absence of globe in the presence of ocular adnexa (eyelids, conjunctiva and lacrimal apparatus). Micropthalmia refers to a globe with a total axial length that is at least two standard deviations below the mean for age.[2]

The birth prevalence of anophthalmia and microphthalmia has been estimated to be 3 and 14 per 100,000 population respectively and the combined birth prevalence of these malformations at up to 30 per 100,000 population. In one –third cases, both microphthalmia and anophthalmia can occur as a part of syndrome.[3]

Anophthalmia/microphthalmia have complex aetiology which includes gestational infections, environmental, monogenic and chromosomal abnormalities. The risk of anopthalmia and micropthamia was significantly higher for live born infants with low birth weights and gestational ages.[4] Of monogenic cause, only SOX2 has been identified as a major causative gene. Other linked genes include PAX6, OTX2, CHX10 and RAX.[5]

Morrison et al[6] conducted a national study including 198 cases of microphthalmia, anophthalmia, and coloboma (MAC) in Scotland and found homozygous loss of function mutations in PAX6 in a single infant with anophthalmia associated with severe brain abnormalities. CHX10 mutations have been described in two families with non-syndromal microphthalmia segregating as an autosomal recessive trait. The SIX3 gene underlies some cases of holoprosencephaly with one patient having microphthalmia and coloboma without other classical features of holoprosencephaly.

Mann[7] suggested that anophthalmia is the result of failure of development of either the anterior neural tube which is known as secondary anophthalmia, or optic pit(s) to enlarge and form optic vesicle(s) which is primary anophthalmia. A third category, consecutive or degenerative anophthalmia was applied to cases where optic vesicles have degenerated and disappeared subsequent to formation.

Fitzpatrick et al suggested that observations of optic nerves, chiasma, and/or tracts with anophthalmia may indicate the regression of a partially developed eye rather than aplasia of the optic vesicle.[8]

Congenital cystic eye is discovered at the time of birth, as a cystic lesion filling the orbit, located behind the upper or lower eyelid without any evidence of eyeball.[9] These cysts are fluid filled lined externally by dense fibrous connective tissue, to which skeletal muscle and adipose tissue are attached. Immature retinal tissue usually lines the inner aspect of the tissue. Because of the developmental failure of the placode, the lens is always absent.[10,11]

Elder[12] found that microphthalmic eyes with a corneal diameter of 5 mm or less at birth were associated with a visual acuity of no perception of light in 81% cases. If doubt persists as to the level of vision then electrodiagnostic tests should be considered.

Diagnosis can be made pre- and post-natally using a combination of clinical features, histopathological study, imaging (ultrasonography and CT/MR scanning) and genetic analysis.

CT and MR scans in anophthalmia show the absence of a globe within the orbit although soft amorphous tissue may be discerned. Neural tissue forming the visual pathway and extraocular muscles are variably present. Orbital volume and dimensions are both reduced. Simple microphthalmia shows as a normal small globe, with normal signal/density characteristics of lens and vitreous, in a smaller orbit than normal.[3]

It is possible to detect anophthalmia/microphthalmia by early second trimester though more recent reports place the limits at about 12 weeks with trans- vaginal ultrasound. Eye size can be measured upon visualizing the maximum coronal or axial planes of the orbit and compared against established eye growth chart.[13]

A study conducted by McLean[14] has outlined the treatment regimen used at Moorfields Eye Hospital which is based primarily on early and aggressive use of conformers. A grossly enlarged cyst was excised soon after presentation and replaced with an orbital implant and ocular prosthesis. Large cysts (with no proptosis from the palpebral fissure) were left to encourage orbital growth. Small cysts (any microphthalmic remnant) were left and the socket treated with aggressive expansion with moulded conformer.

Differential diagnosis from other cystic lesions of the orbit should be based on absence of other recognizable structures in the orbit. So in our case, we had to differentiate between neural cyst of orbit or teratoma or lymphangioma.

In conclusion, although congenital cystic eye is discovered at birth, diagnosis is usually established during surgery demonstrating complete absence of recognizable eye structure. Therapy aims to maximize existing vision and enhance cosmetic appearance rather than improve sight.

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Cite This Article as: *Harakuni U, Smitha KS, Chougule N, Patil M, Sudhakaran P, Linda MS. A Rare Case of Left Sided Anophthalmos with Congenital Cystic Eyeball with Right Sided Microphthalmos.*

Acknowledgments: Nil

Conflict of interest: None declared

Source of Funding: None

Date of Submission: 09 April 2018
Date of Acceptance: 02 May 2018

Address for correspondence

Madhura Patil MS
Girnar Lab, Kolhapur, Sangli Road,
Jaysingpur, Maharashtra - 416101
India
Email id: madhurapatil20.mp@gmail.com

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