

A Rare Case Report of Bilateral Congenital Orbital Colobomatous Cyst with Bilateral Microphthalmos

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Abstract: Colobomatous cyst of the orbit is a rare congenital cystic malformation associated with ocular maldevelopment. Usually, the cyst is associated with a microphthalmic globe. We, herein present a one day old neonate in which parents complained of a bluish mass underlying lower eyelid of both the eyes for which it was evaluated clinically, radiologically and followed up for one year. Cyst aspiration and excision was done later, once cyst growth was stabilised. Histopathology proved to be bilateral large colobomatous cyst associated with bilateral microphthalmos

Key Words: Colobomatous cyst; congenital, microphthalmos, histopathology, aspiration

I. Introduction

Microphthalmos with orbital cyst is a rare congenital cystic abnormality of the globe and orbit that is caused by faulty closure of the posterior part of the embryonic fissure. The cysts project through a congenital defect (coloboma) in the wall of a microphthalmic eye and are lined by a neuroectoderm^[1,2]. It is usually diagnosed at birth and can affect one or both the globes^[2]. This developmental anomaly may appear either as an isolated finding or in association with other ocular and systemic abnormalities^[3]. There is no sex prevalence and in contrast to a similar malformation in animals the majority of cases are non-hereditary^[4].

II. Case report

One-day-old female neonate born out of non-consanguineous marriage presented with swelling of both eyes lower lids since birth. Child was unable to open both the eyes. Right eye examination (Fig 1) showed a diffuse swelling in lower eyelid measuring around 2.5x2cm in size from medial canthus, bluish in colour with regular surface, irreducible and cystic in consistency with no evidence of increase in swelling on crying. Skin over the swelling was pinchable. Left eye examination (Fig 1) showed a diffuse swelling in lower eyelid measuring 2x1cm from medial canthus, bluish in colour with regular surface and cystic in consistency which does not increase on crying. Skin over the swelling was pinchable. Transillumination test was positive in both eye swellings. Both the eyes further ocular examination could not be done because of inability to retract the eyelids. Antenatal and intrapartal history was uneventful. A general paediatric examination revealed otherwise normal healthy child. It was referred to the radiology department with the diagnosis of a bilateral orbital mass.

B-scan Ultrasonography of the right eye showed a cystic lesion measuring 1.5x1.2cm in the inferior quadrant, with no clear communication with the eyeball. Similarly on the left side (Fig 2) cystic lesion measured 1.7x1.4cm with a clear communication to the eye ball through the posterior globe defect. On A-scan, the axial length of the right eye was 10.5 mm and that of the left eye was 8 mm.

On CT examination (Fig 3) of bilateral orbits and brain. There was a large cystic lesion situated in the inferior compartment of the bilateral orbit with superior displacement of the globe. Together, these lesions constitute colobomatous cystic eye disease. The cyst wall was uniformly thin, with a CT density equal to that of normal sclera. The cyst cavity was homogenous, with no enhancement. The globe size was 1x1cm and the size of the cyst was 1.50 x 1.27 cm.

MRI was performed 4 days after birth and revealed (Fig 4) bilateral microphthalmos with a deformed eyeball. Bilateral hyperintense to vitreous cystic lesion was noted on T2 weighted images and hypointense to vitreous on T1W image in the extraconal location, inferotemporal to globe measuring a diameter of 1.5cm on the right and 1.7cm on the left. Communication of cyst with that of the left globe was demonstrated through a defect in the posterior part of the globe.

The patient was diagnosed as a case of bilateral colobomatous cyst with clear communication into the left globe. No obvious pathology was noticed in the brain. No associated malformation noted.

After one year of follow up when cyst remained stable for 6 months, baby underwent an inferior transconjunctival orbitotomy, with aspiration of the contents of the cyst to reduce its volume, followed by excision of the cyst. Ocular prosthesis was fitted to restore cosmesis and to promote growth of the

orbit. Histopathology of the cyst revealed the outer wall consisting of dense fibrocollagenous tissue. The cyst wall was lined with highly vascularized glial tissue .

III. Discussion

Microphthalmos with orbital cyst is a congenital anomaly of the globe caused by a defect in the closure of the embryonic fissure at the 7 to 20-mm stage of development during the 6–7 weeks of gestation^[1]. A majority of the cases of colobomatous cyst are associated with microphthalmos.

The etiology of a colobomatous cyst is not exactly known but it is presumed to occur due to improper fusion of the embryonic fissure. This results in abnormal ectasia of the sclera, which grows into the adjacent orbit^[5]. Usually, the uveal contents do not develop in the region of the coloboma. Shields and associates reported that microphthalmos and colobomatous cyst accounted for 2% of 193 orbital cystic lesions and <1% of 645 biopsies^[6]

The classification of congenital cystic lesions of the orbit encompasses into various subgroups.

- 1) Neural cysts, in which it includes colobomatous cyst associated with ocular maldevelopment,
- 2) A congenital cystic eye, and
- 3) Cysts associated with brain and meningeal tissue (cephalocele and optic nerve meningocele)^[7]

Clinical examination is the key for diagnosis. The presentation of microphthalmos with a cyst can be as a protruding mass in the inferior orbit associated with a malformed microphthalmic eye; The cyst may be so small that it cannot be detected clinically or it may be so large that it obscures the globe^[8]. These eyes usually have a poor visual outcome. Foxman and Cameron^[3] reported that bilateral microphthalmos with colobomatous cyst may be associated with major systemic abnormalities (central nervous system, renal, or cardiovascular), whereas unilateral involvement is usually associated with minor abnormalities. The vision is usually poor. In our case there was no associated abnormalities.

Nowadays imaging techniques as A and B scan ultrasonography^[9] computed tomography^[5] and magnetic resonance imaging^[10] are helpful in supporting the diagnosis and in differentiating it from a congenital cystic eye^[11] meningocele, or meningoencephalocele, primary optic nerve sheath cysts^[12] and teratomas of the orbit. Orbital teratomas quite often have a cystic appearance and are usually benign, although malignant changes have been reported^[13]. Imaging also helps in revealing any communication between the cyst and the globe. This information is useful when planning the management of these lesions.

Management of these cysts depends on the age of the patient, the size of the cyst, the presence of communication between the globe and the cyst, and the visual prognosis^[14].

There is no empirical management of a cyst that may accompany these conditions^[15]. Once the cyst was diagnosed, the goal of management was to keep the cyst to encourage the development of the eyelids and bony orbit. Although surgery may be necessary for some patients, the initial approach for this patient was observation. However, the cyst became enlarged over time. Possible explanations include excessive fluid production by the glial cells lining the cyst wall with pronounced microvilli; communication between the cyst and subarachnoid space; and extensive proliferation of glial tissue that eventually filled and expanded the cyst cavity.^[16-18]

Repeated aspiration of the cyst can be a successful management approach and can be performed with minimal distress to the infant as it was done in our case. In our patient After 1 year of follow-up, the cyst had remained stable for 6 months and later the cyst excision was done. Kodama et al^[19] also reported success with repeated aspiration in patients. The authors recommend that aspiration is considered for patients with extreme colobomatous orbital cyst to maximize the development of the bony orbit.

4. FIGURES



Fig 1: Bilateral bluish subcutaneous mass noted in both eye lower eyelids.



Fig 2:- b mode ultrasound scan of the left eye shows well defined anechoic cystic lesion measuring 1.5x1.2cm inferolateral to the globe with microphthalmos . Note the defect in the posterior part of the globe which communicated with the cyst wall cavity



Fig 3:- Axial contrast enhanced CT of bilateral orbits demonstrates the bilateral cystic lesions within the orbit with no peripheral and internal contrast enhancement. Note the right eye microphthalmos

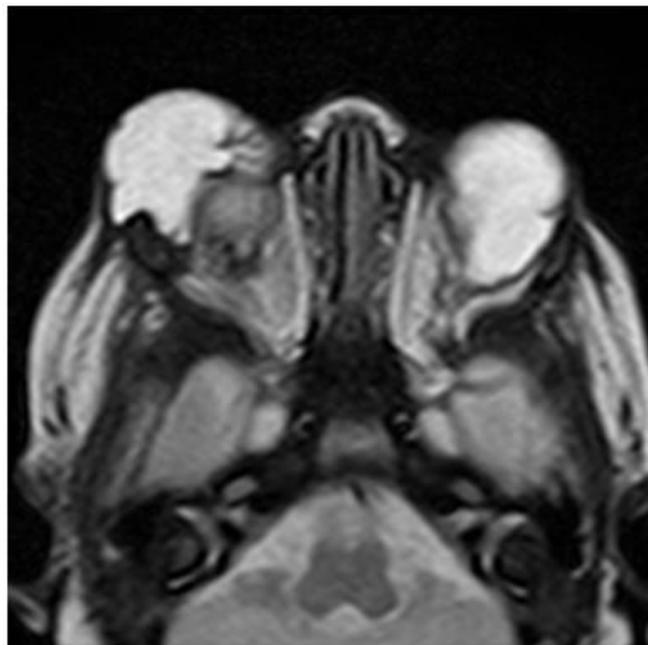


Fig 4:- MRI T2W image depicts homogenous hyperintense well defined pyriform shaped cystic lesion in the bilateral orbit with few hypointense septae within .

IV. conclusion

Bilateral Microphthalmos and colobomatous cyst is a rare, severe developmental anomaly of the globe that results from a defect in closure of the embryonic fissure at the 7- to 20-mm stage of development. Imaging

plays a prime role in differentiating these lesions from other tumors and other cystic lesions like encephalocele etc. Management varies from simple aspiration of the cyst, enucleation of the microphthalmic eye along with the cyst, and excision of the cyst with preservation of the globe depending on the age of the patient and size of cyst. We managed our case with cyst excision which had a reasonable bony orbital growth. These cases are best managed by globe-preservation surgery for the cosmetic purposes.

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