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Congenital left orbital colobomotus cyst associated with micropthalmous in 16 year old male child: Case report

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Abstract
Microphthalmos with orbital cyst is a rare congenital, embryogenic abnormality of fetal fissure closure leading to a small eyeball associated with an orbital cyst. It is accompanied by remarkable repercussions such as cataract, glaucoma, retinal detachment or various ocular positional alterations. It can also lead to other severe facial, neurological, or syndromic anomalies. Observation is debated until the age of 5 when there is no visual threat (i.e. cases with small cysts, slow progression, sight not threatened), in order to prevent orbital underdevelopment. There is no consensus on the optimal technique to remove or reduce the detrimental abnormality. Surgical removal can be considered; either partial exegesis or an aggressive enucleation. The cautious direct aspiration of the cyst is another option with a high recurrence rate. The importance of an early and proper diagnosis and follow-up is well known. Performance of systemic tests, imaging scans, and an electrophysiological study should be considered. We represent the case report describes a rare presentation of a congenital left orbital colobomotous cyst associated with microphthalmia in a 16-year-old male patient.

Keywords: Microphthalmos, colobomotus cyst, orbital cyst, congenital eye disease

Introduction
Congenital microphthalmos is a rare entity with a reported prevalence of 1.4-3.5 per 10000 live births [1]. However, there is no accepted prevalence rate due to the rarity of this disease [2]. Microphthalmos associated with an orbital cyst is even rarer with only few case reports and case series been reported in the literature since it was first described by Arltin 1858 [3].
Microphthalmia is common congenital malformation of the eye after congenital cataract. However, its association with intraorbital cyst is very rare with the prevalence ranging from 1.4 to 3.5/10,000 births and results from failure of closure of optic fissure [4]. It is a neuroectodermal lined cystic mass protruding through coloboma in microphthalmic eye wall. Ocular development begins early in the intrauterine life at 4 weeks with formation of the primary optic vesicle, an outpouching from diencephalon which, then, forms the optic cup with invagination of anterior wall of optic vesicle into posterior wall, while enclosing lens placode derived from surface ectoderm. Retina, iris, and optic nerve are derived from the optic cup, and lens and cornea from the lens placode. Mesoderm grows into choroidal fissure and forms the supporting tissues [5]. Developmental arrest can occur at any stage and the timing of embryological insult decides the presenting clinical condition. Early developmental arrest at 2–7 mm stage before invagination of the primary optic vesicle results in anophthalmos with cyst, whereas a late developmental arrest at 7–14 mm stage results in microphthalmia with cyst as ocular structures have already formed [6].
It appears usually during the first few months after birth and is usually unilateral, though bilateral cases have been reported. It may appear either as an isolated anomaly or in association with other ocular and systemic abnormalities [7].

Case report
16 years old male delivered through normal vaginal Delivery at home and has small eyes with loss of vision since birth and the left eye swelling since 2 years. The swelling had been gradually increasing in size. Swelling was initially painless and now painful since for last 2 months. There were no symptoms related to the cardiopulmonary or gastrointestinal systems. His mother did not attend any antenatal clinic during the pregnancy. He is second child of a 37-year-old housewife and his father is a shopkeeper. There is similar complaints in family member his elder sister is blind since birth. The mother had no history of episodes of febrile illness or ingestion of any drug during the pregnancy. There was a history of delayed developmental milestones. He was not immunized.

Clinical findings
On examination, the patient was found to have:
Left eye: Microphthalmia, with an apparent left orbital cyst extending from the left lower eyelid. The left pupil was poorly formed, and there was no light perception.

Right eye: The right pupil was also poorly formed, and there was no light perception.

Visual acuity: no perception of light in both eyes.

Diagnostic assessment
Imaging
There is evidence of well defined intraconal cyst in left orbit 4x3x4cm with fallowing signals
1. hypointense on T1
2. Hyperintense on T2

Fig 1: Left orbital cyst extending from the left lower eyelid

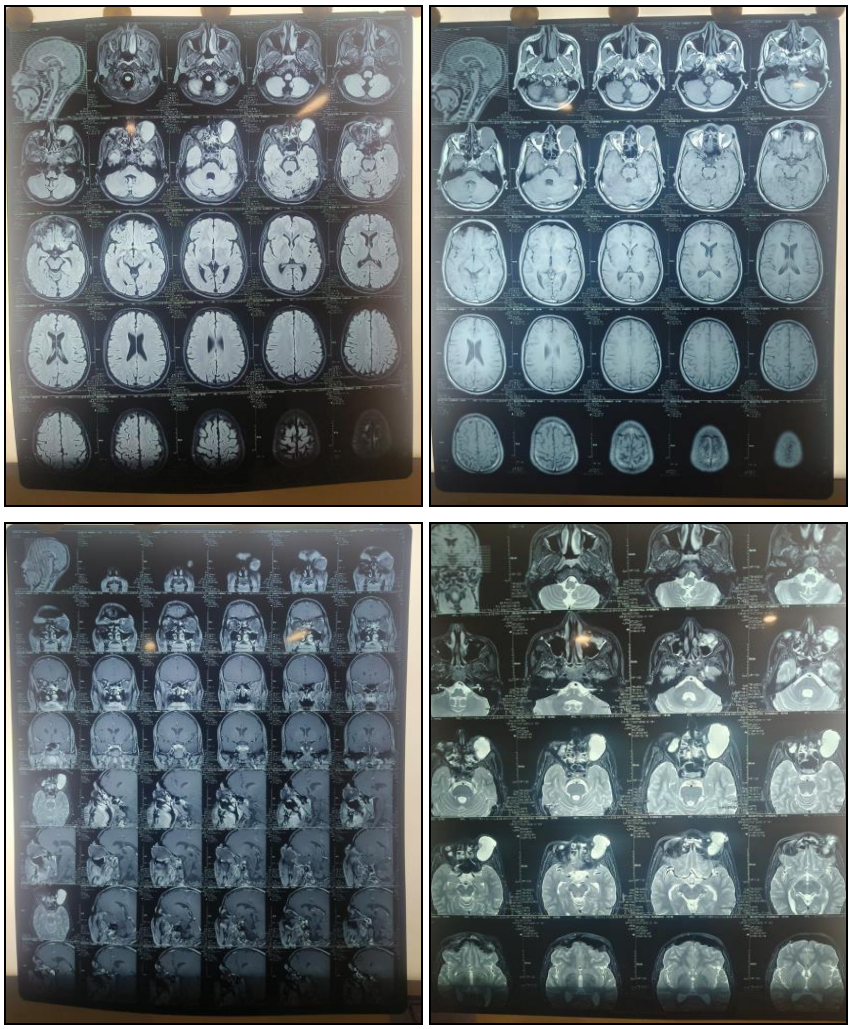

Fig 2: Intraconal cyst in left orbit

Treatment and Management

The patient underwent surgical excision of the colobomotous cyst in the department of neurosurgery at superspeciality Hospital, an associated hospital of Government Medical College Srinagar. The procedure involved:

- A careful approach to minimize trauma to surrounding tissues.
- Postoperative management included antibiotic therapy and regular follow-ups.

Outcome and follow-up

At the 6-month follow-up, the surgical site showed no signs of infection, and the patient reported improved cosmetic appearance and the patient was counseled about the implications of his left eye condition, after that patient has lost follow up.

Discussion

Microphthalmia with cyst is a rare congenital malformation. Orbital cyst with microphthalmos occurs due to defective closure of embryonic fissure at 7–14 mm stage [4]. The entity is usually unilateral. Bilateral involvement, though less common, is more frequently associated with extraocular

malformations [8]. Diagnosis of microphthalmia with cyst is made on clinical basis; however, role of orbital imaging cannot be overemphasized. Imaging assessment is done to evaluate organ of origin, cyst wall, internal contents, and relationship to adjacent structures. USG is quick, noninvasive, and easily available and helps in determining the status of the eye whether anophthalmic or microphthalmic. Computed tomography (CT) depicts the bony abnormalities very well and is employed for presurgical planning with its 3-D reconstruction which is of greater clinical significance in hypoplastic orbit, whereas MRI with its excellent soft-tissue contrast helps characterize the internal contents and wall of the lesion, detect communication channels between the globe and the cyst, and also identify any associated neurological abnormalities [9, 10].

Management is not standard for every case and principally depends on visual potential, cyst size and orbital volume. Simple small cysts with normal ranged orbital volume should be observed while others treated surgically [4]. Surgery is usually recommended before school age for cosmetic appearance, since 90% orbital bone maturation was already completed until school age [11, 12]. Our case was 16 years old male and has small eyes with loss of vision since birth and the left eye swelling since 2 years.

Two unilateral cases (Jensen 1965; Fledelius 1996) and one bilateral case (Ehlers 1966) have previously been published [13, 14, 15]. The previously presented cases of bilateral microphthalmia with cyst, the cysts had made contact with the eyes. Removal of both eye with cyst is the most frequent therapy [16]. Congenital microphthalmos with cyst is defined in three categories, a relatively normal eye with a small cyst, clinically not apparent, an obvious cyst associated with a grossly deformed eye, lastly a large cyst which has pushed the globe backwards so that it is not visible clinically [17]. Severe microphthalmia according to the anatomic appearance and severity of the reduction of the globe refers to a globe with a corneal diameter less than 4 mm and a total axial length less than 10 mm at birth [18]. The present patient was having unilateral presentation of Left Eye: Microphthalmia, with an apparent left orbital cyst extending from the left lower eyelid. The left and right pupils was poorly formed, and there was no light perception in both eyes.

Reviews regarding the management of microphthalmos with colobomatous cysts [19, 20, 21, 22] agree on the therapeutic rationale. It is essential to detect it as early as possible [21]. It allows monitoring to prevent progression towards complications, and to preserve or even improve visual acuity [23, 24]. The main objective is to maintain this visual function. When vision is already irreversibly impaired, the focus is on improving aesthetics and comfort. Some of these reviews [20, 21, 25] emphasize the electrophysiological study, including electroretinogram and visual evoked potentials, to determine this visual potential. Together with imaging tests, which delimit distribution, size and consistency, they are the basis for choosing the most appropriate therapeutic option [20, 21]. All analyses [19, 20, 21, 22] always conclude that the difficulty lies in choosing the most optimal strategy for each individual case. The present patient underwent surgical excision of the colobomotous cyst. A careful approach was used to excise the cyst to minimize trauma to surrounding tissues. Postoperative management included antibiotic therapy and regular follow-ups. At the 6-month follow-up, the surgical site showed no signs of infection, and the patient reported improved cosmetic appearance. Patient was counseled about the implications of his left eye condition.

Conclusion

This case highlights the association between congenital orbital colobomotous cysts and microphthalmia, emphasizing the need for a comprehensive evaluation in such patients. The multidisciplinary approach to management and the importance of patient education regarding potential visual impairment are critical.

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