Microphthalmia with Pseudogliomatosis Proliferation in the Retina and Orbital Cyst

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Abstract

A 28-year-old systemically healthy male presented with a history of decreased vision in the right eye and a blind, undersized left eye since birth. On evaluation, the patient had typical inferonasal iris coloboma and retinochoroidal coloboma in the right eye and left disorganized microphthalmic globe with colobomatous cyst and contracted socket. The patient underwent left eye injection of 0.25 ml ethanolamine olate injected into the cyst after aspiration of the cyst contents. The patient had persistent colobomatous cyst, for which he underwent left eye excision of the microphthalmic globe with orbital cyst. Histopathological examination revealed thickened cyst wall, multiple nodules of dysplastic retinal tissues without proper lamination and extensive surrounding fibrosis, deposition of eosinophilic material around the blood vessels and retina, and multiple foci of calcification without atypia and mitosis. The diagnosis of a unilateral microphthalmic phthisical eye with pseudogliomatosis proliferation in the retina and in the orbital cyst was made.

Keywords: Colobomatous cyst, microphthalmia, pseudogliomatosis proliferation

Introduction

Microphthalmia with orbital cyst is a rare congenital anomaly caused by failure of closure of optic fissure.[1] These patients typically present with a microphthalmic eye with an orbital cyst situated inferior to the globe.[2] They may rarely be associated with gliomatous proliferation. The cysts are lined by neuroepithelium continuous with the vitreous cavity. Microphthalmia with a cyst is usually unilateral. They can be sporadic in origin but may also be associated with systemic anomalies. We herein present a patient with microphthalmia and colobomatous cyst with pseudogliomatosis proliferans.

Case Report

A 28-year-old healthy male presented to us with a history of decreased vision in the right eye and a blind, undersized left eye since birth. According to the patient, his parents were healthy with no history of any congenital malformations in their families, and there was no evidence of consanguinity. On examination, the best-corrected visual acuity was 20/60, and there were J1 (Jaeger) in the right eye and nil light perception in the left eye. The patient had left-sided facial hypoplasia. His right eye showed a microcornea, inferonasal typical iris coloboma, tilted optic disc, Ida Mann Type 3 inferonasal typical retinochoroidal coloboma, and normal intraocular pressure. The left eye showed a microphthalmic globe with an inferiorly located orbital cyst and a contracted socket. The fundus of the left eye was not visible. Systemic examination was normal. Ultrasonography of the left orbit revealed a disorganized globe with grossly reduced axial length, ocular coat calcification, and anterior orbital colobomatous cyst with heterogenous reflectivity inside the cyst [Figure 1]. Prosthesis fitting was not possible in the left socket.

The patient underwent left eye aspiration of the cyst contents followed by transconjunctival injection of 0.25 ml ethanolamine olate. However, the orbital cyst persisted with there being no response to medical treatment. So, the left eye was excised. Histopathological examination revealed thickened cyst wall, multiple nodules of dysplastic retinal tissues without proper lamination and extensive surrounding fibrosis, deposition of eosinophilic material around the blood vessels and retina, and multiple foci of calcification without atypia and mitosis. The diagnosis of a unilateral microphthalmic phthisical eye with pseudogliomatosis proliferation in the retina and in the orbital cyst was made.

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inability to fit prosthesis in the left socket. Computerized tomography (CT) scan of the orbit revealed a rudimentary left globe displaced superotemporally and a lobulated isodense colobomatous cyst with specks of calcification seen within the cyst [Figure 2].

The patient underwent left eye excision of the microphthalmic globe along with the orbital cyst measuring 24 mm anteroposteriorly, 22 mm horizontally, and 18 mm vertically. Histopathological examination revealed thickened cyst wall, multiple nodules of dysplastic retinal tissues without proper lamination and extensive surrounding fibrosis, multiple dilated thickened blood vessels, deposition of eosinophilic material around the blood vessels and retina, multiple pseudorosettes, suggestive of dysplastic retinal tissue, bone formation by the retinal pigment epithelial cells, and multiple foci of calcification. The eyeball specimen consisted of necrotic intraocular contents with possible bone formation and phthisical changes. There was no atypia or mitotic activity [Figure 3].

The diagnosis of a microphthalmic phthisical eye with pseudogliomatosis proliferation in the retina and in the orbital cyst was made.

**Discussion**

Microphthalmia and coloboma with orbital cyst are rare congenital anomalies arising during 6th–7th week of gestation due to defects in closure of embryonic fissure and invagination of optic vesicle, resulting in the varied clinical presentation of microphthalmia with cyst.[1,2] The orbit volume is usually reduced due to microphthalmia. The mechanism of cyst formation is poorly understood. It is postulated to result from prolapse of intraocular contents through a scleral defect. Another explanation is that microphthalmia with cyst represents ectasia of the colobomatous globe.[3,4] The diagnosis is usually made during the first few days of life. This condition is usually isolated and can be either unilateral or bilateral.[1]

Association with systemic anomalies may occur. Sporadic cases may be associated with the Goldenhar, CHARGE, or VATER syndrome.[5] It may also be due to teratogenic agents, maternal ingestion of drugs, infection, fever, or irradiation. In these cases, possibility of abnormal neural crest cell development has been suggested.[4]
Neuroepithelial elements such as astrocytes, oligodendroglia, and microglia are present in the retina. The neurogia of the brain and retina is well-known to be able to proliferate under the influence of irritants. Glial reaction is a characteristic feature in the eyes marked by chronic inflammation.[5] Thus, congenital pseudogliomatous proliferation in the presence of a coloboma can cause propagation of the tissue through the fissure with formation of cysts in the orbit. Progressive enlargement of the cyst may be seen due to extensive proliferation of glial tissue, eventually filling and expanding the cyst cavity.[6]

The diagnosis is based on oculo-orbital B-mode ultrasound and CT and/or magnetic resonance imaging scan to assess the state of the eyeball and the orbit. CT scan is better modality to demonstrate the eyeball and the cyst walls, as well as any communication between the two.[4,7]

Orbitopalpebral cysts are usually associated with a deformed globe with poor visual prognosis. Small colobomatous cysts will be respected, with regular monitoring. Unsightly bulky cysts may be excised with repair of the orbital cavity.[4] Cosmetic rehabilitation often requires excision or aspiration of the cyst to allow space for prosthesis. Aspiration of the cyst is often preferred over surgical cyst excision which often leaves behind a contracted socket as unwanted sequelae to surgery. However, simple cyst aspiration is frequently associated with recurrences. Recurrence can be avoided using a sclerosing agent such as ethanolamine oleate.[4] Surgical options include placement of orbital implants of fixed dimensions at one or more surgeries or use of a dermis-fat graft which has the capability of postsurgical growth [Figure 4].[9,10]

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has his consent for his images and other clinical information to be reported in the journal. The patient understand that his name and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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