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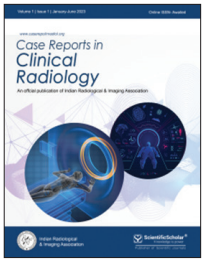

Case Report

Congenital ocular cyst with microphthalmia – A case report

Renu Yadav[1], Anju Ranga[1], Mahesh Kumar Mittal[2], Mukul Sinha[1]

[1]Department of Radiodiagnosis, Vardhaman Mahavir Medical College and Safdarjung Hospital, New Delhi, [2]Netaji Subhash Chandra Bose Subharti Medical College, Meerut, Uttar Pradesh, India.

***Corresponding author:**
Renu Yadav,
Assistant Professor, Department of Radiodiagnosis, Vardhaman Mahavir Medical College and Safdarjung Hospital, New Delhi 110029, India.

docrenu0803@gmail.com

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ABSTRACT

Congenital microphthalmia with intraorbital cyst is rare. Colobomatous cyst stems from defect in closure of embryonic fissure at the time of invagination of optic vesicle, leading to protrusion of neuroectodermal lined cystic mass from the defect in the wall of microphthalmic eye. Clinical presentation usually comprises progressively enlarging orbital cyst with microphthalmia. Enlargement of orbital cyst is attributable to continuous secretions from its wall. Imaging assessment evaluates lesion and its internal characteristics, organ of origin, and associated anomalies. Differentials include microphthalmia with cystic teratoma and meningoencephalocele. Management strategy comprises removal of cystic lesion or aspiration, followed by sclerotherapy with bleomycin injection and ocular expander to maintain orbital volume for prosthesis.

Keywords: Microphthalmia, Colobomatous cyst, Ocular dermoid cyst, Venolymphatic malformation

INTRODUCTION

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.[1] 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.[2] 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.[3]

CASE REPORT

A 27-year-old male patient presented with complaints of progressively increasing right orbital swelling since birth with complete loss of vision. No history of trauma could be illicited. No previous imaging evaluation was done. On local examination, the right orbit appeared larger than the left with a tense swelling involving entire orbit which was non-tender to touch and showed

very good transillumination. Clinically, no globe could be visualized. The eyelids were normal. The left eyeball was normal with normal visual acuity. Clinical diagnosis of the right side congenital cystic eye with anophthalmia was made.

Ultrasonography showed enlarged right orbit occupied by cystic lesion with few septations within. In superomedial part of orbit, a small solid area resembling a small residual eyeball was seen with hyperechoic areas, small calcific foci, and vascularity on color Doppler [Figure 1].

Magnetic resonance imaging (MRI) revealed a large trans-spatial cystic mass lesion involving both intra- and extraconal compartment measuring approximately 5.5 × 3.4 × 4.9 cm (AP × Trans × CC). The lesion was hypointense on T1WI and hyperintense on T2WI with multiple T2 hypointense septations of variable thickness, maximum thickness measuring approximately 3 mm. No evidence of any hemorrhage or calcification was noted within the cystic areas. Solid T2 hypointense area was seen in superomedial portion, showing diffusion restriction on DWI, blooming on Gradient echo sequences (GRE), and homogeneous post-contrast enhancement. The right optic nerve was atrophied and seen coursing until the solid area in superomedial part, likely dysgenetic eye [Figure 2]. Lesion had led to marked widening of orbital fossa with bony remodeling.

Computed tomography was done for better evaluation of the bone again confirmed the ultrasound (USG) and MRI findings and showed cystic lesion with microphthalmia. No evidence of bony erosion/destruction is shown in [Figure 3].

The patient was treated with aspiration of cystic contents. Approximately 40 mL of clear straw colored fluid was aspirated followed by bleomycin injection into the collapsed cyst to prevent reaccumulation [Figure 4].

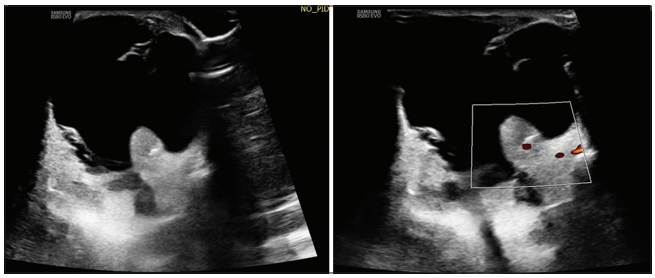

Figure 1: Sonographic image of the patient with color Doppler shows multiloculated cystic lesion with thin internal septation and eccentric solid area with focus of calcification, showing color flow on color Doppler imaging in superomedial compartment.

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.[1] The entity is usually unilateral. Bilateral involvement, though less common, is more frequently associated with extraocular malformations.[4] 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, non-invasive, and easily available and helps in determining the

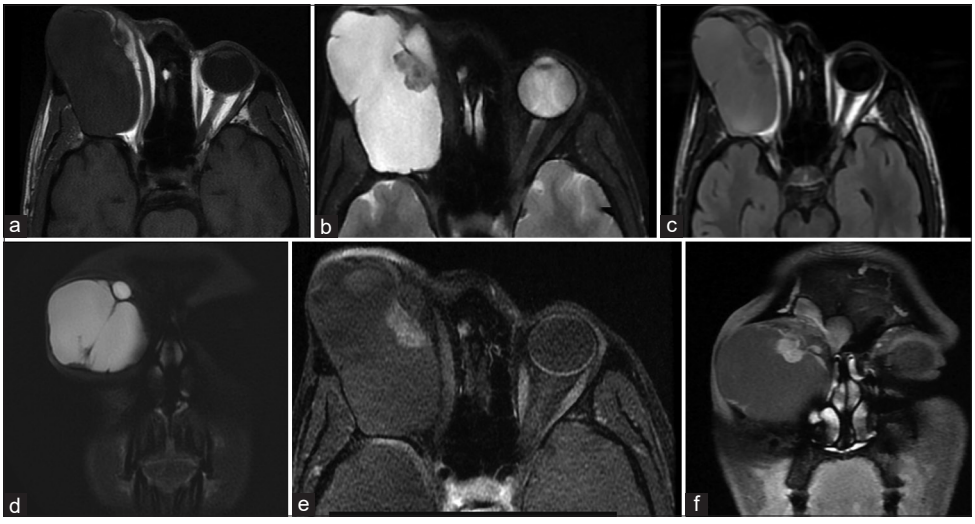

Figure 2: Magnetic resonance imaging T1 Axial (a), T2 axial (b), T2 Flair (c), T2 Coronal (d), and T1 post-contrast axial and coronal (e and f) show a multicompartmental cystic lesion with thin septations within which appears hypointense on T1, hyperintense on T2, incomplete suppression on Flair, enhancement of thin septae on post-contrast imaging along with solid enhancing area and microphthalmic eye in superomedial compartment and T2 hypointense tract extending from microphthalmic eye to cyst (d).

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.[5,6]

Imaging findings include a well-defined multicompartment cystic lesion extending into both intra- and extraconal compartments with few septations that may show vascularity or contrast enhancement on CT/MRI. No evidence of any fat/calcification/bleed/phlebolith is seen within the lesion. Eccentric enhancing solid component with calcific foci may be seen in the superomedial compartment of orbit representing rudimentary globe. Communication between globe and cyst may or may not be seen.[7] A thinned out and atrophic optic nerve can be traced in most cases from the solid rudimentary globe. The bony orbit may be enlarged with remodeling.

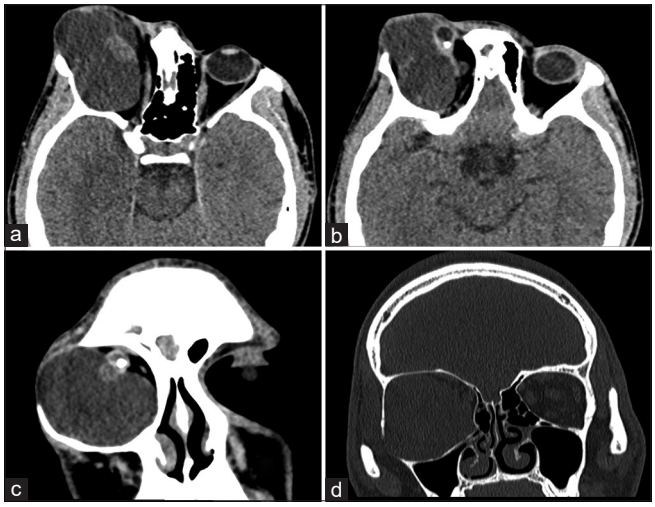

Figure 3: Axial (a and b) NCCT and coronal (c) image soft-tissue window show cystic lesion with few thin internal septations within and eccentric solid soft-tissue resembling globe seen in superomedial part of orbit. NCCT coronal image (d) shows expansion of bony orbit with wall thinning compared to the left side.

Histopathological examination demonstrates a cavity lined by inner primitive neuroretinal layer and outer layer of fibrovascular connective tissue. There is no evidence of any mitosis, nuclear atypia, or necrosis.[8]

Differential diagnoses include orbital cystic teratoma, encephalocele, congenital cystic eye, and venolymphatic malformation.[5] None of the above-described lesions are associated with microphthalmos.

Orbital dermoid cysts are the most common orbital masses in children, are frequently extraconal, and smoothly marginated cystic lesions with areas of fat/calcification.[5] Meningocele or encephalocele are intraorbital herniation of meninges and/or brain parenchyma. Intracranial communication of cyst can be depicted well with imaging. In congenital cystic eye, the entire globe is replaced by cyst with non-visualization of globe.[5] Venolymphatic malformation clinically does not show brilliant transilluminance as shown by cyst due to its lymphatic contents, presents with progressively enlarging orbit, or may have acute presentation with hemorrhage into one of the loculations. Furthermore, venolymphatic malformations are unencapsulated, diffusely infiltrative with multicompartment involvement and show fluid-fluid levels on MRI.[9]

Once a clinicoradiological diagnosis of microphthalmia with cyst is made, the next step is to evaluate for visual potential and any other associated anomalies which are more common if disease is bilateral. Orbit attains 90% of adult size by 6 years of age; hence, cyst in a child of <7 years should be left untouched considering its contribution to orbital growth. If treated earlier than this age, close monitoring for the orbital growth and volume using expander needs to be ensured for proper future prosthesis implantation. This approach aims to minimize asymmetry and achieve desired esthetic and functional outcomes. Complete surgical excision of the cyst with orbital prosthesis is the ideal treatment method. Aspiration of the cyst alone is avoided due to high rates of

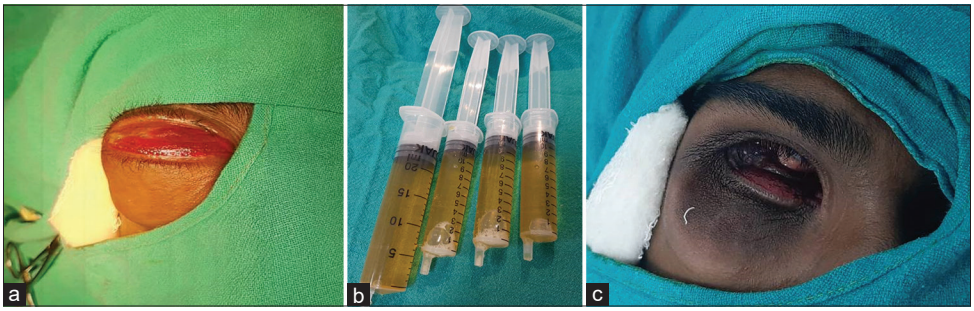

Figure 4: Clinical image of the patient preoperatively (a), around 45 mL of clear straw colored fluid (b) was aspirated and bleomycin was injected, then post-operative eye showing microphthalmic eye in superomedial part (c).

re-accumulation of fluid. Concomitant sclerotherapy helps to incite fibroinflammatory response within cyst cavity and avoid relapse.

CONCLUSION

Colobomatous cyst with micropthalmia is benign, developmental lesion presenting as progressive increase in eye ball size. Optimal management is aspiration with concomittent destruction of wall lining to prevent reaccumulation followed by prosthetic eye placement

TEACHING POINTS

1. Microphthalmia with cyst is a rare benign congenital malformation
2. It is differential diagnosis of more commoner lesions like orbital cystic teratoma, encephalocele, congenital cystic eye, and venolymphatic malformation
3. Clinicoimaging evaluation can accurately diagnose disease
4. Management strategy is aspiration with concomitant sclerotherapy to prevent reaccumulation followed prosthetic eye replacement.

MCQs

1. True about colobomatous cyst are all except -
   a. Congenital pathology
   b. Due to faulty embryonic development
   c. Can be associated with anophthalmia or microphthalmia
   d. If left untreated can undergo malignant change

   Answer Key: d
2. Differential diagnosis of congenital cystic are all except-
   a. Orbital teratoma with microphthalmia
   b. Meningocoele or encephalocoele
   c. Retinoblastoma
   d. Colobomatous cyst with microphthalmia

   Answer Key: c
3. True about management of colobomatous cyst are-
   a. Aspiration
   b. Aspiration followed by sclerogent injection and prosthetic eye replacement
   c. Can be left alone
   d. Resection

   Answer Key: b

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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