

# Bilateral microphthalmos and unilateral colobomatous orbital cyst: successful management by repeated aspiration

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## Abstract

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. This report presents the clinical features and outcome of an infant with bilateral microphthalmos and a right colobomatous cyst treated with repeated aspiration.

**Key words:** Coloboma, Cyst, Infant, Microphthalmos

## Introduction

In a study of more than 50,000 pregnancies in the USA, the incidence of anophthalmia and/or microphthalmia was 0.22 per 1000 births and the prevalence among blind children was between 0.6% and 1.9%.<sup>1</sup> 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.<sup>1</sup> Fluid accumulation within the cyst can cause it to extend in front of the eye remnants and protrude through the eyelids leading to exposure, poor cosmesis, and infection.<sup>2</sup> Microphthalmos is usually diagnosed soon after birth and can affect one or both globes. This developmental anomaly may appear either as an isolated finding or in association with other ocular and systemic abnormalities.<sup>3</sup>

The clinical presentation may vary and could lead to misdiagnosis. Imaging studies such as ultrasonography,

computed tomography (CT), and magnetic resonance imaging (MRI) are helpful for making the diagnosis.<sup>4</sup> Although management is usually conservative, extremely large cysts may be treated either by repeated aspiration or surgical removal of the cyst. Due to the communication of the cyst with the eye, excision may deflate the microphthalmic eye resulting in removal of the eyeball.

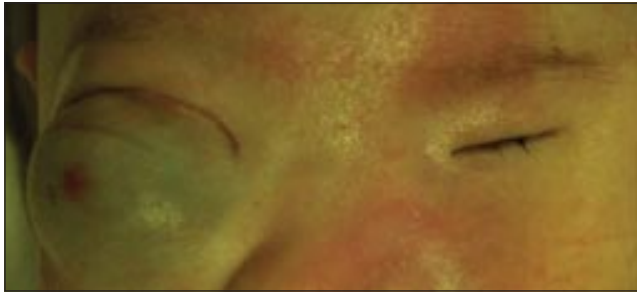
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## Case report

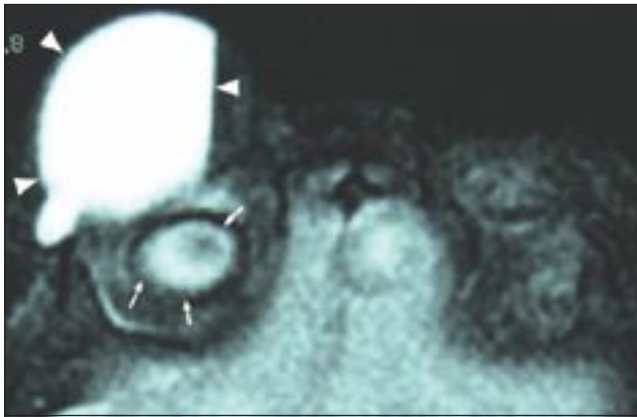
A male infant weighing 3.11 kg was born at 40 weeks' gestation with bilateral microphthalmos and a right colobomatous cyst. His parents showed no consanguinity, and there was no history of birth defects or drug ingestion during pregnancy. Chromosomal analysis showed normal karyotyping. Neither parent had any ocular abnormality.

External examination of the right eye showed a microphthalmic globe with a hazy microcornea that was displaced superonasally by a large transilluminated firm subcutaneous bluish mass occupying the inferotemporal part of the orbit. Fundusoscopic examination could not be performed because of the cloudiness of the cornea. External examination of the left eye revealed extreme blepharophimosis with no identifiable ocular tissue in the conjunctival sac (**Figure 1**).

Other craniofacial abnormalities included occipital encephalomeningocele, schizencephaly, dilated ventricular



**Figure 1.** A 1-month-old neonate with left clinical anophthalmos and right microphthalmos with a large orbital cyst.



**Figure 2.** Magnetic resonance imaging of the orbit shows right microphthalmos (arrows) with a cyst (arrowheads) showing hyperintense signals in T2-weighted image.

system, small posterior fossa, and left complete cleft lip and palate. Other major systems were all normal.

Ultrasound examination of the right eye showed a microphthalmic globe filled with medium reflective tissue and a large echolucent cystic structure extending to the orbital apex. On the left side, an extreme microphthalmic globe without any cystic structure was found. MRI was performed 5 days after birth and revealed no identifiable globe over the left socket and a deformed right eyeball with a cyst in the extraconal location of the anterior orbit with a diameter of 2.5 cm. The cyst was covered with skin and was located inferior and anterior to the globe. No communication between the ventricular system and the cyst was demonstrated (Figure 2).

By the time the infant was 9 weeks old, the cyst had markedly increased in size and completely obscured the microphthalmic globe. The cyst was aspirated in sterile conditions under general anesthesia — 7 mL of brownish fluid was obtained. A maximum size conformer was inserted. Cytology revealed mainly hemosiderin-laden macrophages and red blood cells and was negative for malignant cells. However, the fluid rapidly reaccumulated and the aspiration process was repeated under local anesthesia 5 more times: at 10 weeks, 6 mL of brownish fluid was aspirated; at 13 weeks, 7 mL was aspirated; at 15 weeks, 3 mL was aspirated; at 19 weeks, 3 mL was aspirated; and at 23 weeks, 2 mL was aspirated. After 23 weeks, the cyst had not refilled and remained deflated at 1 year (Figure 3). An



**Figure 3.** After 1 year of follow-up, the right orbital cyst regressed after repeated aspiration with residual soft tissue swelling.



**Figure 4.** Computed tomography scan of the orbit shows right microphthalmos (arrowheads) with the regressed orbital cyst (arrows).

ocular prosthesis was fitted to restore cosmesis and to promote growth of the orbit.

CT scan of the orbit performed when the infant was 8 months old revealed bilateral microphthalmos with severe optic nerve hypoplasia. A soft tissue density of 0.8 cm in diameter was located posterior and inferior to the right globe. The mass was connected to the apex of the right orbit but was not connected to the right optic nerve or right inferior rectus muscle (Figure 4).

## Discussion

Differential diagnosis of congenital cystic orbital masses includes encephaloceles, dermoid cyst, arachnoid cyst, congenital cystic eye, and solid tumor with central necrosis. These conditions may all present with similar clinical and imaging findings.<sup>4</sup> However, these lesions are not associated with microphthalmos. Microphthalmos due to other causes such as persistent hyperplastic primary vitreous, retrolental fibroplasias, congenital infectious ophthalmopathy, and nanophthalmos are not accompanied by an orbital cyst.<sup>5</sup>

Foxman and Cameron reported that bilateral microphthalmos and colobomatous cyst can be associated with major systemic abnormalities (central nervous system, renal, or cardiovascular), whereas unilateral microphthalmos and colobomatous cyst may be associated with only minor abnormalities.<sup>3</sup> This patient had multiple craniofacial abnormalities (occipital encephalomeningocele, schizencephaly, dilated ventricular system, bilateral microphthalmia with right orbital cyst, small posterior fossa, and cleft lip and

palate). However, this range of abnormalities does not apply to any specific syndrome.

There is no empirical management of a cyst that may accompany these conditions.<sup>6</sup> Once the cyst was diagnosed, the aim of management was to keep the cyst for as long as necessary 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, completely obscuring the microphthalmic globe. 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.<sup>7-9</sup>

This report demonstrates that repeated aspiration of the cyst can be a successful management approach and can be performed with minimal distress to the infant. After 1 year

of follow-up, the cyst had remained stable for 6 months. However, it is crucial to confirm by MRI or CT that no communication exists between the cyst and any intracranial structures such as the ventricular system or encephalocele before performing aspiration. Kodama et al<sup>10</sup> and Mathew and Peter<sup>2</sup> also reported success with repeated aspiration in 2 separate patients. In the patient in our report, the volume of fluid accumulating within the cyst was less following each aspiration, and physical laws may explain this phenomenon — the Law of Laplace states that the pressure required to keep a thin-walled hollow structure expanded is directly proportional to the tension in its wall divided by its diameter.<sup>11</sup> Therefore, greater effort is required to initiate the inflation of a cyst than to increase the volume of a larger cyst.

The authors recommend that aspiration is considered for patients with extreme colobomatous orbital cyst to achieve possible globe preservation and maximize the development of the bony orbit.

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