

**Case Report****Congenital Bilateral Cryptophthalmos with Orbito-Palpebral Cyst and Microphthalmos**

**Mohammad Sadegh Dehghani Firouzabadi <sup>1</sup>, MD; Shima Alsadat Dehghanpour Farashah <sup>\*1</sup> MD; Afsaneh Yarmohammadi <sup>2</sup>, MS, MD**

1- Geriatric Ophthalmology Research Center, Shahid Sadoughi University of Medical Sciences, Yazd, Iran.

2- Rajaei Cardiovascular, Medical and Research Institute, Tehran, Iran.

**\*Corresponding Author:** Shima Alsadat Dehghanpour Farashah

**E-mail:** ms\_dehghani\_f@yahoo.com

**Abstract**

Microphthalmia is regarded as the most common congenital malformation of the eye, second only to congenital cataract. Its association with an intraorbital cyst, however, is exceedingly rare. Cryptophthalmos represents a rare anomaly characterized by the partial or complete absence of the eyebrow, palpebral fissure, eyelashes, and conjunctiva. In this condition, the partially developed adnexa are fused to the anterior segment of the globe, leading to severe ocular defects. In this report, we present a rare case of congenital bilateral cryptophthalmos accompanied by an orbitopalpebral cyst and microphthalmos and the treatment results for the patient.

**Keywords:** Anophthalmia; Microphthalmia; Cryptophthalmos; Congenital; Orbito-Palpebral Cyst.

**Article Notes:** Received: Jun. 14, 2022; Received in revised form: Jul. 03, 2022; Accepted: Jul. 27, 2022; Available Online: Jan. 01, 2023.

**How to cite this article:** Dehghani Firouzabadi MS, Dehghanpour Farashah SA, Yarmohammadi A. Congenital Bilateral Cryptophthalmos with Orbito-Palpebral Cyst and Microphthalmos. *Journal of Ophthalmic and Optometric Sciences*. 2023;7(1): 24-27.

## Introduction

Cryptophthalmos is an extremely rare ocular disorder that may occur in isolation or as part of Fraser syndrome<sup>1</sup>. Eyes with cryptophthalmos represent a fundamental failure in ocular development and have a very poor prognosis for visual function<sup>2</sup>. Francois classified cryptophthalmos into three subtypes: complete, incomplete, and abortive forms<sup>3</sup>. In most cases of complete cryptophthalmos, the primary objective of surgical intervention is to improve cosmetic appearance, since there is no visual potential<sup>3</sup>.

Congenital microphthalmia is a rare unilateral or bilateral disease, characterized by a decreased size of the eyeball and orbit. It can present independently or as part of syndromes associated with congenital malformations in other organ systems<sup>4</sup>.

The aim of this case report is to present the clinical observations and the reconstructive procedure that achieved satisfactory cosmetic results in a rare patient with bilateral cryptophthalmos, orbitopalpebral cyst, and microphthalmia.

## Case Report

A term newborn girl was presented in our labor room with the chief complaints of being unable to open both eyes, accompanied by swelling beneath both lower eyelids since birth, without any other anomalies (Figure 1). We received written consent from the parents to present this case report.

The patient was otherwise healthy and had no family history of a similar developmental anomaly. The mother, 18 years old, and the father, 23 years old, were first-time parents with no previous history of abortion. The mother had no history of illness, trauma, or



**Figure 1:** The patient presented with closed eyes, accompanied by swelling beneath both lower eyelids

exposure to toxic agents or radiation during pregnancy.

It appeared that the infant had no light perception in either eye. Clinical evaluation revealed bilateral cryptophthalmos and enlarged globes of differing sizes, both of which were of soft consistency. Ultrasonography revealed microphthalmos and an acoustically hollow lesion consistent with an associated cyst in both orbits. A Computed Tomography (CT) scan of the orbits showed dysplastic globes characterized by cystic expansions that extended laterally, anterior to the lateral orbital rim (Figure 2).

Based on the clinical and imaging findings, the diagnosis was bilateral cryptophthalmos with orbitopalpebral cyst and microphthalmos, presenting a poor cosmetic appearance. The cysts on both sides were removed, and the fornix of both eyes was reconstructed; subsequently, a conformer was inserted into both eyes to improve the cosmetic appearance (Figure 3).



**Figure 2:** Dysplastic globes characterized by cystic expansions that extended laterally, anterior to the lateral orbital rim



**Figure 3:** The patient after reconstructive surgery and insertion of a conformer in both eyes

## Discussion

Cryptophthalmos, derived from the Greek meaning “hidden eye,” was first introduced by

Zehender, who described a pediatric patient with eyes completely covered by skin<sup>3</sup>.

Egr1, a critical zinc finger transcription factor for differentiation, plays a vital role in mammalian eyelid development and closure, influencing ocular integrity<sup>5</sup>. A deficiency in Egr1 can lead to abnormal eyelid development and closure, predisposing individuals to a variety of ocular abnormalities, including cryptophthalmos<sup>5</sup>. Moreover, exogenous factors such as a history of viral infection, drug, or toxic gas exposure during early pregnancy may contribute to the etiology of cryptophthalmos<sup>6</sup>. Rehabilitating cryptophthalmos presents a challenging scenario, and the primary surgical goal is to restore cosmetic appearance and salvage visual potential, although, in most complete cryptophthalmic cases, there is no visual potential<sup>3</sup>.

Microphthalmia is the most common congenital malformation of the eye after congenital cataract<sup>7</sup>. Unilateral microphthalmia is more common than bilateral, with only one-third of cases being bilateral<sup>7</sup>.

Our patient presented with bilateral cryptophthalmos, orbitopalpebral cyst and microphthalmos, and had a poor cosmetic appearance requiring surgery. The orbital ultrasound B and CT scans were instrumental in diagnosing the condition, aiding not only in diagnosis but also in identifying other abnormalities. It was an isolated malformation without systemic defects.

Chaudhry et al.,<sup>8</sup> have recommended that in severe microphthalmia, both the cyst and globe should be excised, and volume replaced. In our case, due to the large cyst and rudimentary microphthalmic eye, enucleation, cyst excision, fornix reconstruction, and conformer insertion were performed to improve cosmetic appearance.

## Conclusion


Here, we reported a rare case of congenital bilateral cryptophthalmos with an orbitopalpebral cyst and microphthalmos. The surgical management involved enucleation, cyst excision, fornix reconstruction, and conformer insertion to improve cosmetic appearance.

## Authors ORCIDs

Mohammad Sadeq Dehghani Firouzabadi:

 <https://orcid.org/0000-0002-5434-0954>

Shima Alsadat Dehghanpour Farashah:

 <https://orcid.org/0009-0005-4125-5610>

## References

- 1- Farooqui JH, Thiên Hà TH, Gomaa A. Isolated Bilateral Complete Cryptophthalmos. *Nepal J Ophthalmol*. 2016;8(16):186-8.
- 2- Saleh GM, Hussain B, Verity DH, Collin JR. A surgical strategy for the correction of Fraser syndrome cryptophthalmos. *Ophthalmology*. 2009;116(9):1707-12.
- 3- Liu Z, Xie B, Li Y, Ding J, Li D. Reconstruction strategy in isolated complete Cryptophthalmos: a case series. *BMC Ophthalmol*. 2019;19(1):165.
- 4- Gittens A, Gedrich M, Khandelwal M, Fischer R, Germaine P. Isolated presentation of congenital microphthalmia on fetal MRI. *Radiol Case Rep*. 2019;14(9):1069-71.
- 5- Oh J, Wang Y, Chen S, Li P, Du N, Yu ZX, et al. Genetic background-dependent role of *Egr1* for eyelid development. *Proc Natl Acad Sci U S A*. 2017;114(34):E7131-9.
- 6- Ding J, Hou Z, Li Y, Lu N, Li D. Eyelid and fornix reconstruction in abortive cryptophthalmos: a single-center experience over 12 years. *Eye (Lond)*. 2017;31(11):1576-81.
- 7- Timalisina BR, Shrestha GB, Thapa M. Congenital Microphthalmia with Intraorbital Cyst: A Rare Case Report. *Case Rep Ophthalmol Med*. 2019;2019:3640175.
- 8- Chaudhry IA, Arat YO, Shamsi FA, Boniuk M. Congenital microphthalmos with orbital cysts: distinct diagnostic features and management. *Ophthalmic Plast Reconstr Surg*. 2004;20(6):452-7.

## Footnotes and Financial Disclosures

### Conflict of Interest:

The authors have no conflict of interest with the subject matter of the present study.