Acta Scientific Ophthalmology (ISSN: 2582-3191)

Volume 7 Issue 9 September 2024

Case Report

Non Surgical Approach in the Management of Microphthalmia Associated with a Giant Colobomatous Orbital Cyst; A Case Report

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DOI: 10.31080/ASOP.2024.07.0794

Received: July 25, 2024

Published: August 23, 2024

Abstract

Microphthalmia associated with a colobomatous cyst is a rare embryogenic anomaly. 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. 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.

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. Nevertheless, the possibility of combining it with ethanolamine oleate sclerotherapy can be appropriate in some cases.

We report the case of a newly arrived Moroccan child. He was diagnosed at the age of 13 years with left microphthalmia associated with a giant cyst, which was protruding from the lower eyelid. The scan revealed ocular dysgenic, debris with calcifications, continued by an expansive colobomatous cyst. Given the null visual potential, the option of excision and orbitotomies was contemplated, but a less aggressive approach using ethanolamine oleate sclerotherapy was chosen with good results both in reducing the size of the cyst and in aesthetic and functional improvement of the patient.

Keywords: Microphthalmia; Microphthalmos; Nanophthalmos; Anophthalmos; Colobomatous Cyst; Coloboma, Orbital Cyst; Orbital Anomaly; Congenital Eye Disease; Ethanolamine Oleate; Oleate of Ethanolamine; Sclerotherapy

Introduction

Microphthalmos is due to abnormal conformation of the optic fissure during the sixth and seventh week of embryonic development [1]. There are two main subtypes: simple, in which all ocular structures are adequately functional, and complex, involving abnormal structures such as iris coloboma or colobomatous cysts [2]. The term 'coloboma' refers to an incomplete closure of either the uvea (iris coloboma) and/or the neuroectoderm with subsequent cystic proliferation (chorioretinal colobomatous cyst) [3-7].

Nowadays, this is a pathology with few references in the literature and no evidence regarding its management. Previous publications were focused on describing isolated case reports, acknowledgement of its pathology, and debating its management between observation, surgery or aspiration.

Ethanolamine is a bifunctional molecule containing both a primary amine and a primary alcohol. Ethanolamine functions as a sclerosing agent, used for sclerotherapy of vascular lesions such as

esophageal and rectal varices, and congenital vascular malformations or cysts. It mainly works by irritating the cyst, leading to the formation of fibrous tissue and to the closure of the cyst cavity.

We report a case of successful reduction of a colobomatous cyst related to microphthalmos with the use of ethanolamine.

Case Report

A 13-year-old boy from Morocco consulted for a left ocular deformity since birth, which caused him discomfort and made it impossible for him to see. The patient had no pathological history of interest, nor any other systemic anomaly.

On examination, the visual acuity of the right eye was 100% but the left eye could not perceive light. The right orbit (RO) had a normal ocular anatomy, meanwhile the left orbit (LO) was bigger in size and had a lower lid mechanical ectropion. The lower eyelid was everted due to a large whitish protruding mass occupying all the orbit, the contents of which were unknown. There was no prior acknowledgement of the existence of ocular remnants.

A computed tomography (CT) scan was performed to clarify the diagnosis and rule out urgent orbital complications, which demonstrated a severe microphthalmos continuing from a large inferolateral intraorbital cyst (Figure 1). It confirmed that there was an absence of viable ocular structures and even remodeling of the orbital floor. Visual evoked potentials and electroretinogram showed totally abolished visual capacity. The physical, analytical, genetic and imaging study at the systemic level was unremarkable.

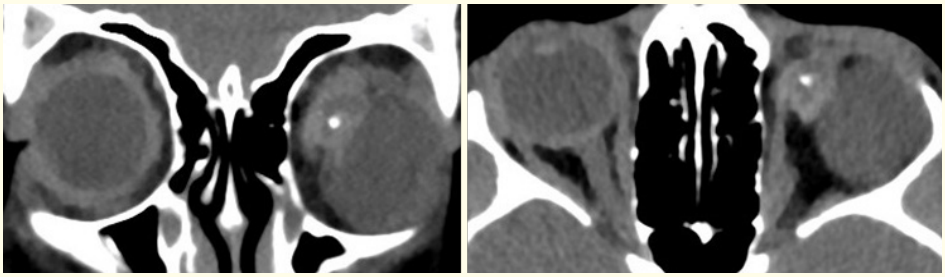

Figure 1: Coronal (A) and axial (B) orbital CT scans; dysgenic left microphthalmia with calcifications in its interior, continuing from a cyst measuring 23 x 28 x 28 millimeters, probably colobomatous. There was also optic nerve atrophy (B).

Given the lack of vision and the very advanced situation, it was considered that the classic surgical option would end up being very aggressive and would even require several surgeries. For this reason, one of the alternatives proposed was sclerotherapy with ethanolamine oleate.

We proceeded to perform this procedure. In the supine position, with general anesthesia and a blepharostat, we exposed the cyst. With the help of a 25-gauge needle, we aspirated 4 ml of cyst fluid. This way most of the fluid was removed. Afterwards we injected 2 ml of ethanolamine oleate at a dilution of 1.25% into the hollow cyst. With a single course of therapy, the objective was achieved. The cyst contracted sufficiently to stop protruding the eyelid and a cosmetic shell could be adapted. The posterior analysis of the aspirate corroborated the colobomatous origin. We prescribed antibiotic plus corticosteroids ointment for the next few days and the child did not complain of significant short- or long-term side effects.

The 6-month post-injection CT scan revealed this significant decrease in the large left colobomatous cyst (Figure 2). One-year post-injection, the good result was maintained at follow-up, with no further procedures required.

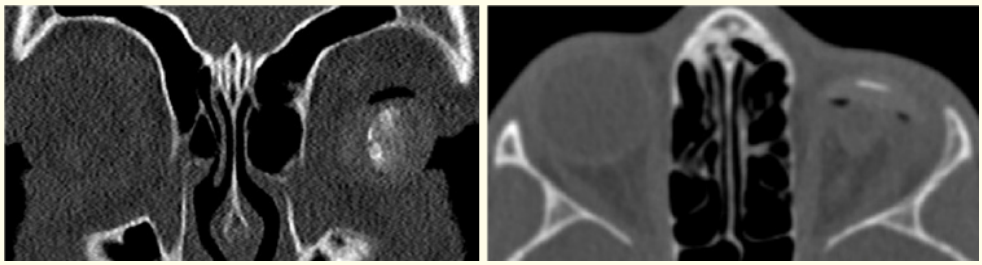

Figure 2: Coronal (A) and axial (B) orbital CT scans; significant reduction of the cyst, remaining minimal dysgenic-calcified cyst remnants behind. There was remodeling of the orbital floor.

Discussion and Conclusion

Microphthalmia associated with a colobomatous cyst is one of the rarest congenital ocular anomalies, with an incidence of only 1 in 10,000 births [2,8,9]. It is usually sporadic in possible association with intrauterine infections (such as rubella, varicella, toxoplasmosis, herpes simplex and cytomegalovirus) [10]. However, hereditary cases are also well known [9,11]. Epidemiologically, almost 30% of cases are bilateral [5,6,8,12], and more than 60% have other ocular abnormalities, either in the same eye or in the contralateral eye; angle-closure glaucoma, early cataracts, retinal detachment in the context of chorioretinal coloboma, strabismus and/or proptosis. In addition, half of them have other systemic disorders, especially facial and neurological [9,13-15], or even form part of syndromes, such as Charge's or Gorlin's [10,13,16]. Interestingly, they are found twice as often in males, and the most common location of the cyst, whether or not it is connected to the optic disc, is the infero-nasal region of the eye [15].

In most cases the diagnosis is clinical at birth. It relies on ultrasound as the initial test and magnetic resonance imaging (MRI) as the most conclusive [4,6,9,17]. It is crucial to add a systemic pediatric evaluation. Depending on the context, examination of first-degree relatives, genetic screening and counseling may also be useful [10].

Reviews regarding the management of microphthalmos with colobomatous cysts [2,4-6,9,10,12,18] agree on the therapeutic rationale. It is essential to detect it as early as possible [10]. It allows monitoring to prevent progression towards complications, and to preserve or even improve visual acuity [7,19]. 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 [5,10,18,20] 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 [5,6]. All analyses [2,4-6,9,10,12,18] always conclude that the difficulty lies in choosing the most optimal strategy for each individual case.

In orbits that are still conforming, either with or without vision, both the eyeball and the cyst play an important role in stimulating faco-orbital growth [19-21]. There is scientific consensus to recommend retention of the cyst only until 5 years of age [10,21-23], by which time periocular bone formation is already 90% of the final adult condition. Early removal may aggravate asymmetry and deformity, as well as complicate orbital prosthetic accommodation [22,23]. However, delaying action on the cyst may lead to compression and even ocular underdevelopment. Before the age of 5 years, observation may be most appropriate in a small, slow-growing cyst that does not threaten visual ability.

Although, if insufficient orbital volume is anticipated (axial length of less than 16 mm), hydrophilic conformers of increasing size can be used to stimulate adequate orbital growth [21]. In severe microphthalmia with residual vision, translucent expanders can be used at the visual axis level. However, it is cautioned that in moderate or mild microphthalmos, clear transparency in front of the cornea (with a free visual axis) should always be preferred to prioritize functional benefit [10].

However, if the cyst grows too fast, acquires a risky size, causes deprivation amblyopia or is unfavorable, the first step is to try to remove the cyst in isolation.

Direct aspiration of the cystic contents is the simplest but has the highest recurrence rate; if its capsule does not collapse completely it reaccumulates fluid [21]. Still, it is a safer option when there is visual potential [24,25]. In contrast, simple surgical excision of the cyst, with or without fatty tissue [20], is more definitive, but preservation of vision can be more challenging. Consideration must be given to: the size and density of the cyst, its proximity to the brain, vitreous detachment, and, above all, the point of communication of the cyst with the eye and its extent. It is more difficult to achieve good optical results when wide pedicles have to be ligated and it is impossible to preserve vision if pedicles with a nerve connection have to be cut [21,24-26].

More recently, sclerotherapy by injecting ethanolamine oleate has emerged. Although its use in bleeding gastro-oesophageal varices is better known [27], good references have recently increased in vascular [28,29] and cystic lesions [30-32]. It offers more effective and permanent results than simple aspiration. At the same time, it is more conservative (although less definitive) than surgery [27,33,34]. The only caveat is that it is contraindicated in eyes with visual potential.

A few case reports, detailing this procedure in orbital cysts, were found [33-35]. They avoided topical anesthesia when children were involved. They used 21- or 23-gauge needles and ethanolamine oleate at a dilution of 3 or 5%. They aspirated a variable cyst amount from 2 to 10 ml and they usually injected an amount of ethanolamine oleate of half the one that was aspirated. They all reported important and permanent reductions of the volume cysts.

On the other hand, the classic invasive option is surgical removal of the cyst with the ocular bulb [22,23].

It is indicated in extremely large cysts or in complex anatomies, when the eye also lacks visual potential. Evisceration is not always resolute, but is less invasive than enucleation [12]. Both options carry additional risks: post-orbitotomy infection, and exposure or extrusion of the implant [21]. As well as expandable conformers, expandable implants, either intraconical or intrascleral, can be considered in this context [21].

In a second stage, most patients require ocular prosthesis; clear ones maximize vision in mild or moderate microphthalmos, while painted ones are indicated in severe microphthalmia or anophthalmia cavities [10].

We presented an atypical case; a very rare congenital disease that did not consult until a very advanced stage at 13 years of age due to limited medical resources in his country of origin.

The review of therapeutic options and our preference for the less traumatic ethanolamine injection highlights the need for a clinical and therapeutic guideline that incorporates less invasive non-surgical therapies with a correct risk-benefit balance.

Acknowledgements

This study adhered to the Principles of the Declaration of Helsinki. There was no funding or grant support.

Conflict of Interest

The authors have no financial disclosures.

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