Radiology Case Reports 19 (2024) 5346–5349

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journal homepage: www.elsevier.com/locate/radcr

Case Report

Congenital right optic nerve colobomatous cyst associated with microphthalmos ☆

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ARTICLE INFO

Article history:
Received 28 July 2024
Accepted 31 July 2024

Keywords:
Optical nerve
Coloboma
Orbit
MRI

ABSTRACT

Optic nerve coloboma is a congenital defect caused by the incomplete closure of the embryonic fissure. This closure begins around the fifth week of gestation, when the embryo measures approximately 7 to 14 mm. Colobomas may appear as isolated defects or alongside other ocular and systemic abnormalities. They typically occur in the inferonasal quadrant, where the optic vesicle undergoes closure. This closure process starts centrally within the eye and progresses both anteriorly and posteriorly. The distinct nature of these closures can result in an optic nerve coloboma without an associated iris coloboma.

The formation of a coloboma can be attributed to external factors affecting the fetus during pregnancy or to a genetic mutation, such as an autosomal dominant mutation in the Pax gene, which is linked to bilateral colobomas. Small colobomas generally require only clinical and radiological observation, whereas larger colobomas may need surgical intervention to remove the cyst and prevent complications related to orbital content development, thereby improving cosmetic outcomes

Introduction

Optic nerve coloboma is a congenital defect caused by the incomplete closure of the embryonic fissure. This closure begins around the fifth week of gestation, when the embryo measures approximately 7 to 14 mm [1–3]. Colobomas may appear as isolated defects or alongside other ocular and systemic abnormalities. They typically occur in the inferonasal quadrant, where the optic vesicle undergoes closure. This closure process starts centrally within the eye and progresses both anteriorly and posteriorly. The distinct nature of these closures can result in an optic nerve coloboma without an associated iris coloboma.

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☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2024.07.186

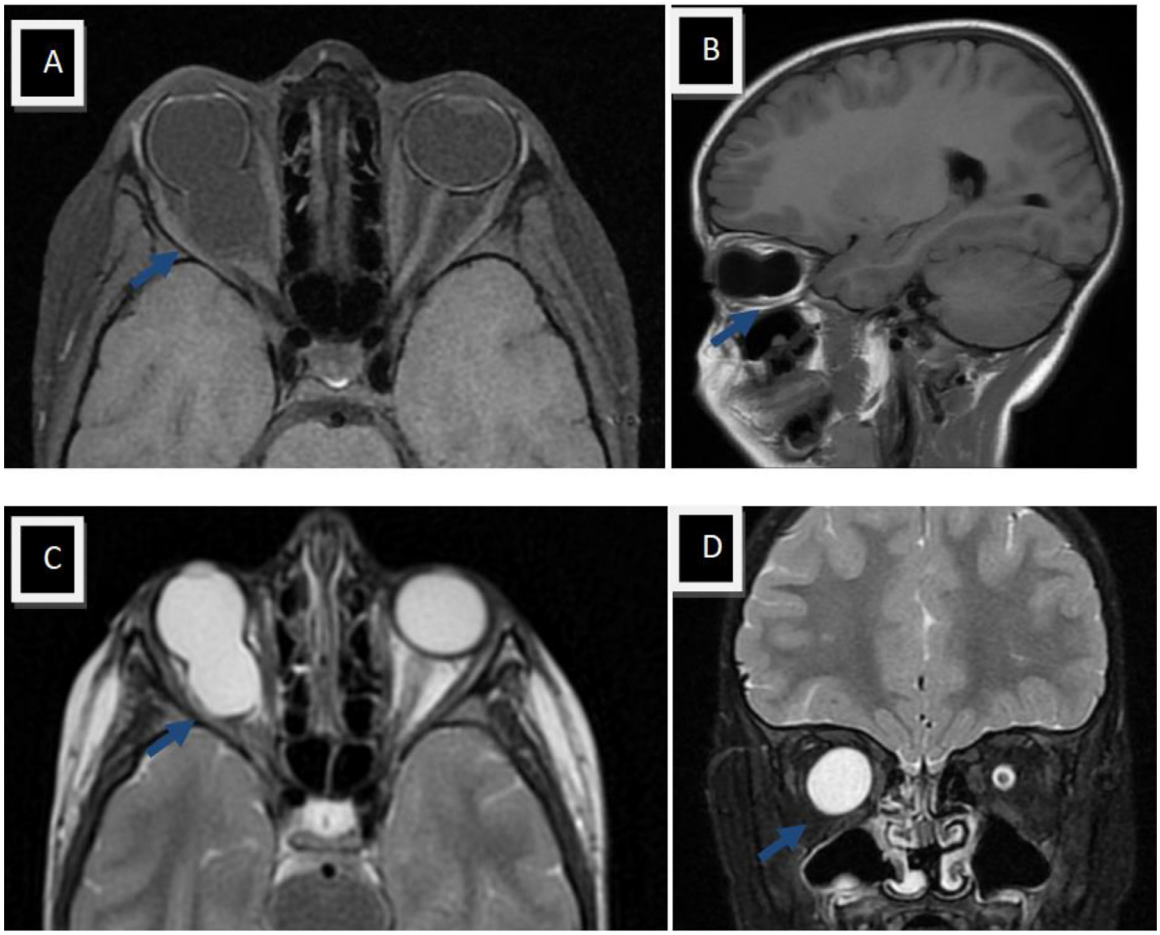

Fig. 1 – (A) Axial T1W, (B) sagittal T1W, (C) Axial T2W, and (D) coronal T2W MRI image, showing identified a T1 hypointense and T2 hyperintense lesion, corresponding to a retrobulbar, intra-conal cystic cavity that completely replaced the optic nerve (blue arrow).

intervention to remove the cyst and prevent complications related to orbital content development, thereby improving cosmetic outcomes [4,5].

Case report

We present the case of a 3-year-old male child with no medical or surgical history, who presented with decreased vision in the right eye. Clinical examination revealed right enophthalmos. Magnetic resonance imaging (MRI) (Fig. 1) identified a T1 hypointense and T2 hyperintense lesion, corresponding to a retrobulbar, intra-conal cystic cavity that completely replaced the optic nerve. Surgical treatment was performed using a lateral Kronlein approach, allowing for the en bloc removal of a large cyst, with the pathological optic nerve ligated at both ends. The cyst contained clear, high-pressure fluid. Histopathological examination revealed neuroretinal tissue surrounded by glial tissue. Skin closure with sutures facilitated good healing of the surgical wound. Three months postoperatively, clinical follow-up showed good eyelid and ocular motility. A follow-up CT scan confirmed satisfactory results, indicating reduced ocular protrusion and the presence of a small residual cavity.

Discussion

Microphthalmos and colobomatous cysts represent rare yet significant developmental abnormalities of the orbit. These conditions result from the incomplete closure of the optic fissure during fetal development, leading to the herniation of glial tissue into the orbital cavity, forming a cystic structure [6]. The affected eyes often exhibit severely impaired vision or even blindness, as observed in the current case where the patient had no vision in the right eye [7].

Microphthalmos with a cystic lesion can present as an isolated congenital defect, as in the current patient, or alongside a spectrum of systemic or intracranial anomalies. It is not uncommon for the contralateral eye to also display coloboma, although in this instance, the left eye appeared unaffected. This diversity in presentation necessitates a careful differential diagnosis to distinguish between colobomatous cysts and other congenital cystic orbital masses. Conditions that may present

with similar clinical and imaging features include encephaloceles, arachnoid cysts, dermoid cysts, and solid tumors with central necrosis. However, these entities are generally not associated with microphthalmos [7–9].

Other causes of microphthalmos that do not involve orbital cysts include persistent hyperplastic primary vitreous, retrolental fibroplasia, congenital infectious ophthalmopathy, and anophthalmos. These conditions can sometimes present with overlapping clinical features, making accurate diagnosis challenging [9].

The clinical presentation of microphthalmos with colobomatous cysts can vary widely, complicating the diagnostic process. Symptoms may range from the presence of a visible cystic mass to more subtle signs of ocular maldevelopment. Advanced imaging techniques, such as magnetic resonance imaging (MRI), computed tomography (CT), and ultrasonography, are indispensable in achieving a precise diagnosis. These imaging modalities are crucial not only for identifying the presence and extent of the cyst but also for detecting any communication between the cyst and the globe or the subarachnoid space/ventricular system. Such communications can influence the choice of treatment and management strategies [10–12].

The management of microphthalmos with colobomatous cysts requires a multidisciplinary approach, involving ophthalmologists, radiologists, and plastic surgeons. Early diagnosis and intervention are critical for optimizing outcomes. The therapeutic approach for colobomatous cystic lesions is multifaceted and must be tailored to the specific circumstances of each patient. Key factors influencing treatment decisions include the patient's age, the size and growth potential of the cyst, the presence of communication between the cyst and the globe, and the overall visual prognosis [13]. The primary objectives of treatment are to promote normal orbital growth and achieve satisfactory cosmetic outcomes. Various treatment options are available, ranging from conservative management with regular monitoring to more invasive surgical interventions. These options include:

- Simple Cyst Aspiration: This approach may be considered in cases where the cyst does not significantly impact the globe or orbital structure. It involves draining the cystic fluid to reduce its size.
- Enucleation of the Microphthalmic Eye with Cyst Removal: This more invasive procedure may be necessary when the cyst is large or causing significant orbital distortion. Enucleation is often followed by orbital reconstruction to support normal growth and improve cosmetic appearance.
- Cyst Excision with Globe Preservation: In cases where preserving the eye is feasible, surgical removal of the cyst while maintaining the integrity of the globe is preferred. This approach aims to retain as much ocular function as possible while addressing the cyst.
- Orbital Expanders and Conformers: These devices can be used postoperatively to encourage orbital development and maintain proper eyelid positioning.

Whenever feasible, globe-preserving surgery is the preferred approach, as it tends to yield better cosmetic results and preserves any residual visual function. The choice of intervention must be carefully weighed against the potential risks, including the possibility of postoperative complications or the need for additional surgeries.

In the case of significant cystic formations, particularly those presenting late, as in the current patient, more extensive surgical intervention may be required. This can pose additional challenges in terms of achieving satisfactory cosmetic rehabilitation and preventing complications such as infection or recurrent cyst formation.

Postoperative follow-up is crucial for monitoring the patient's recovery and for early detection of any complications or recurrence of the cyst. Additionally, regular follow-up is necessary to assess the potential for malignant transformation, although this is relatively rare.

Conclusions

This case report details an uncommon presentation of a congenital colobomatous cyst in the right orbit, accompanied by microphthalmos, which was identified later than typically expected. Utilizing advanced imaging technologies like MRI can greatly aid in the early detection of such lesions, providing valuable information for parental counseling and optimizing early treatment strategies.

Author contributions

All authors contributed to this work. All authors have read and approved the final version of the manuscript.

Guarantor of submission

The corresponding author is the guarantor of submission.

Patient consent

Written informed consent for publication was obtained from patient.

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