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Birth Defects of the Child Optic Disc: Diagnostic Challenges and Prospects for Care

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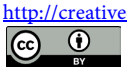

Abstract

Introduction: Congenital optic disc anomalies in children refer to structural variations of the optic nerve head present from birth. These deformations involve the size, shape, color, and vessels of the optic disc. Although often asymptomatic, these anomalies can impact the visual development of the child, underscoring the importance of a thorough fundus examination for early detection and appropriate medical follow-up. We present two cases of congenital optic disc anomalies in children, illustrating the diagnostic challenges and complexity of their management. **Case 1:** A 3-year-old girl presented with a white spot in her left eye present since birth. Uncorrected distance visual acuity was 2/10 in the right eye, while she could perceive hand movements at 2 meters with the left eye. Normal examination in the right eye showed leukocoria, microphthalmia, and a white mass at the center of the optic disc on fundus examination in the left eye. Ocular imaging, including ultrasound and optical coherence tomography (OCT), confirmed the diagnosis of persistent hyperplastic primary vitreous (PHPV) in its mixed form in the left eye. Management included prescribing full optical correction and functional rehabilitation, without resorting to surgery. The course was marked by persistent amblyopia leading to a poor prognosis. **Case 2:** A 7-year-old girl consulted for vision disturbance in her right eye. Visual acuity was finger counting at 2 meters in the right eye and 10/10 in the left eye. Anterior segment examination revealed no abnormalities in both eyes. However, fundus examination highlighted a large funnel-shaped excavation associated with central glial proliferation, wheel spoke vessels, and neuroretinal ring atrophy

in the right eye. Optical coherence tomography (OCT) of the right eye confirmed the diagnosis of isolated unilateral Morning Glory syndrome. Management included full optical correction and orthoptic rehabilitation. The course was marked by the absence of ocular complication and maintenance of visual stability in the right eye. The prognosis seemed favorable. **Conclusion:** Congenital optic disc anomalies in children exhibit great clinical variability and require an individualized diagnostic and therapeutic approach.

Keywords

Optic Disc, PHPV, Morning Glory, Diagnosis, Treatment

1. Introduction

Congenital optic disc anomalies represent a complex area of pediatric ophthalmology, where the clinical variability of presentations and the diversity of manifestations pose diagnostic and therapeutic challenges [1]. They encompass all structural malformations of the optic disc and surrounding tissues, which can lead to congenital visual impairment or even blindness [2]. These structural variations involve the size, shape, color, and vessels of the optic nerve head [3] [4] [5] [6]. Although rare, these conditions can have a significant impact on the visual development of children, requiring differentiated and individualized management. We present two cases of congenital optic disc anomalies that highlight the diagnostic challenges and therapeutic options to preserve ocular health and improve the visual quality of life for affected children.

2. Observations

2.1. Case 1

A 3-year-old girl presented with a white spot in her left eye, present since birth. Visual acuity was 2/10 in the right eye and hand motion at 2 meters in the left eye. Normal anterior segment examination in the right eye revealed leukocoria and microphthalmia in the left eye. Fundus examination was normal in the right eye but showed a white mass extending from the center of the optic disc to the temporal region in the left eye (**Figure 1**). B-mode ocular ultrasound (**Figure 2**) confirmed the presence of a vitreous mass and microphthalmia with an axial length of 21.7 mm. In the face of diagnostic doubts, color Doppler ultrasound (**Figure 3**) showed vascular flow in the retroocular mass. Magnetic resonance imaging (**Figure 4**) confirmed the diagnosis of persistent hyperplastic primary vitreous in its mixed form in the left eye. Management involved prescribing full optical correction and functional rehabilitation to address amblyopia, with no surgical indication. The course was marked by profound amblyopia in the left eye, without anatomical complications. The prognosis for the left eye was poor. This case highlights the complexity of managing PHPV, with an emphasis on the

need for a multidisciplinary approach.

2.2. Case 2

A 7-year-old girl presented with vision impairment in her right eye. Visual acuity was finger counting at 2 meters in the right eye and 10/10 in the left eye. Ophthalmic examination revealed a funnel-shaped excavation at the optic disc in the right eye accompanied by glial proliferation centered on the fine vessels in radial distribution and atrophy of the neuroretinal ring (Figure 5). Optical coherence tomography (Figure 6) ruled out any morphological abnormalities of the optic nerve head or cranioencephalic region and diagnosed isolated unilateral Morning Glory syndrome. Management included full optical correction and orthoptic functional rehabilitation. Ophthalmic follow-up over one year monitored potential changes in the optic disc, visual acuity progression, and the onset of potential complications. The prognosis appears favorable, characterized by the absence of ocular complications and maintenance of visual stability in the right eye. This observation allowed assessment of the psychosocial impact and ensured optimal management of this congenital malformation.

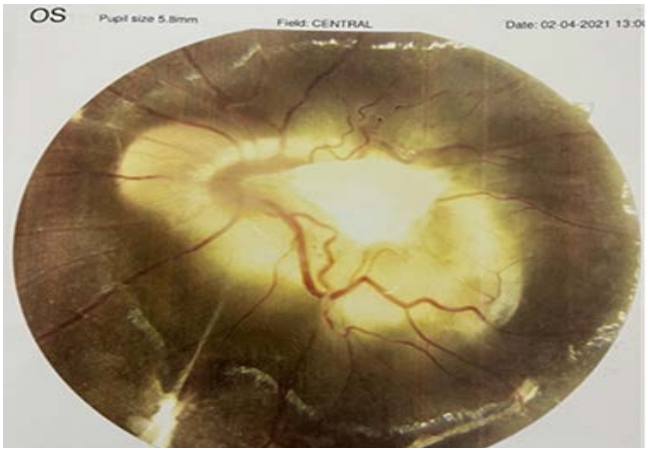

Figure 1. Residual embryonic vitreous in the optic disc of the left eye.

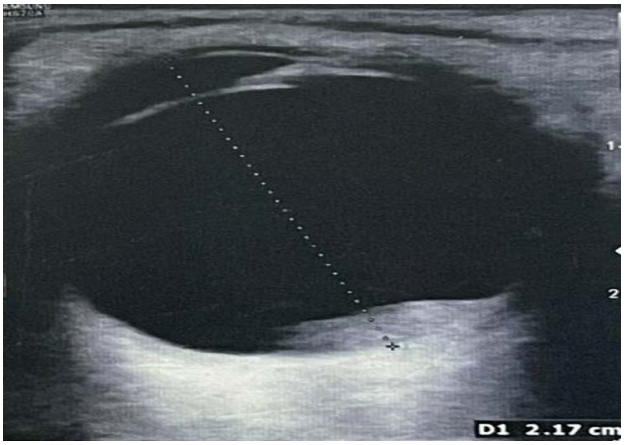

Figure 2. Vitreous in the form of a mass on B-mode ultrasound of the left eye.

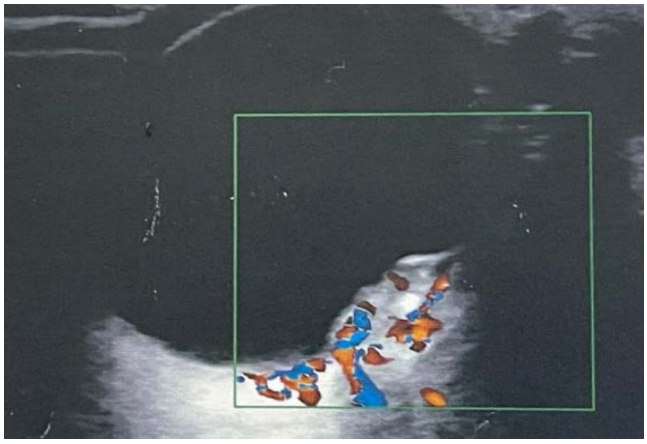

Figure 3. Vascular flow on Doppler ultrasound of the left eye.

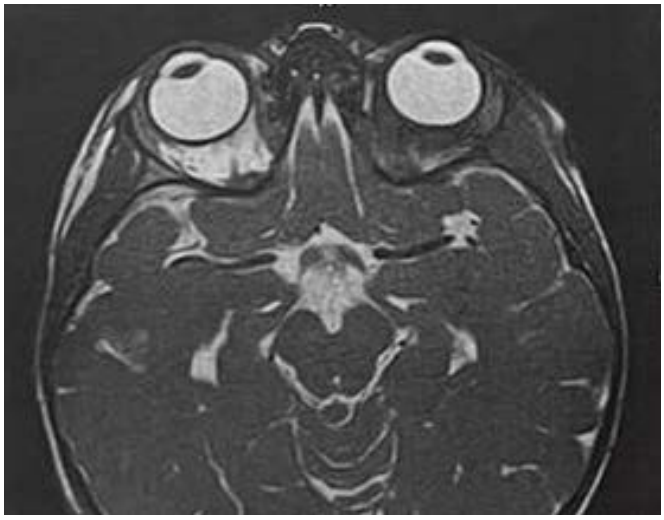

Figure 4. Mixed form of PHPV on MRI of the left eye.

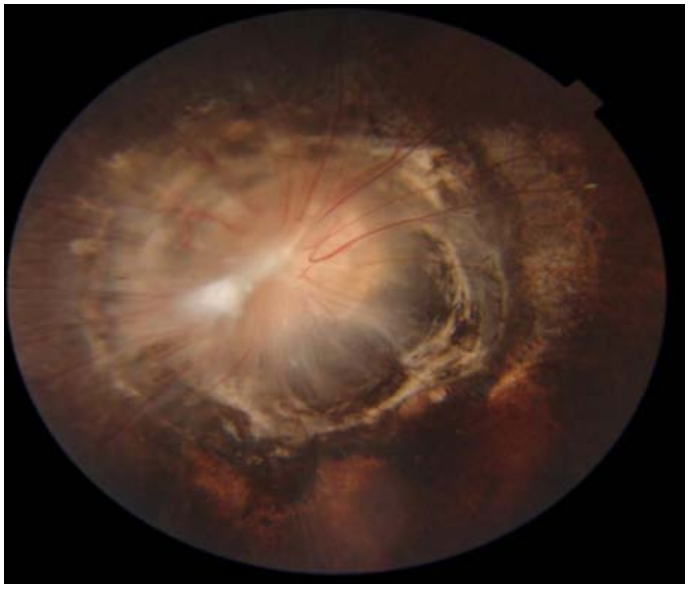

Figure 5. Morning Glory syndrome in the right eye.

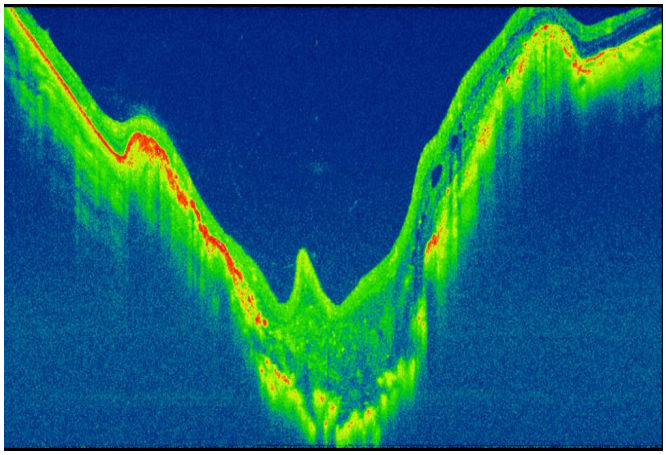

Figure 6. OCT cross-section of Morning Glory Syndrome in the right eye.

3. Discussion

Congenital optic disc anomalies pose a significant challenge in terms of diagnosis and management due to their clinical variability. The cases presented in this clinical observation highlight this diversity, thus illustrating the complexity and the need for a differentiated approach for their evaluation and treatment.

In the first case, the persistence of the hyaloid membrane in its mixed form was diagnosed in a 3-year-old girl, presenting with leukocoria, microphthalmia, and persistent amblyopia in the left eye. Early detection of this anomaly requires meticulous investigation of the optic disc to promptly identify persistent vascular membranes and other structural abnormalities. The complexity of persistent hyperplastic primary vitreous (PHPV) cases is accentuated by clinical variations, necessitating various diagnostic approaches and advanced imaging techniques in addition to thorough clinical evaluation. Many authors emphasize the importance of carefully analyzing the fundus examination to avoid missing these rare conditions in children [5] [6] [7] [8].

However, the second case involved a 7-year-old girl with isolated unilateral Morning Glory syndrome, characterized by a large funnel-shaped excavation at the optic nerve head, without amblyopia, thus having a more favorable prognosis. Diagnosing this syndrome poses diagnostic challenges due to its rarity and variations in clinical manifestations. Various authors have conducted studies confirming the variety of clinical manifestations of this pathology, thereby highlighting the crucial importance of a meticulous fundus examination to avoid missing this diagnosis [9]-[16]. Identifying this congenital anomaly requires precise clinical expertise, with the presence of a funnel-shaped excavation at the optic disc being the specific diagnostic criterion.

A differentiated approach is necessary to understand the subtleties of each case and address the diagnostic challenges specific to each condition. The diversity of presentations underscores the importance of an individualized approach in diagnosing and managing congenital optic disc anomalies.

Technological advancements, particularly in medical imaging, are becoming

essential for diagnosis. Ultrasound, optical coherence tomography (OCT), and magnetic resonance imaging (MRI) provide detailed information, facilitating more precise diagnostic approaches and optimal management of optic disc anomalies in children. These technological advancements offer new opportunities to overcome diagnostic challenges.

However, despite the similar use of advanced diagnostic methods, fundamental distinctions in mechanisms and clinical manifestations reinforce the importance of adopting a specific approach for each condition. By integrating these technological advancements with a differentiated and individualized approach, healthcare professionals can improve diagnostic accuracy and management of congenital optic disc anomalies. Thorough ophthalmological examination in children is essential to differentiate between persistent hyperplastic primary vitreous (PHPV) and Morning Glory syndrome (MGS), involving careful clinical observation and additional examinations.

Congenital optic disc anomalies in children can have a significant impact on their visual development. The optic disc is a crucial structure in the eye where the optic nerve emerges and enters the retina, so any structural variation affecting the size, shape, color, and vessels of the optic disc can compromise the transmission of visual signals to the brain.

In the cases presented, these anomalies were associated with various symptoms, including decreased visual acuity, leukocoria, and microphthalmia. These symptoms can lead to vision impairment and visual development issues in children. The persistent amblyopia observed in one of the children underscores the importance of early intervention to minimize long-term complications.

Thus, early detection is crucial for preserving visual development. Regular eye examinations from early childhood can help promptly identify these anomalies and initiate appropriate management. Therapeutic management has also been individualized, with the prescription of optical correction and the implementation of functional rehabilitation tailored to each case. Additionally, regular medical follow-up is necessary to monitor the progression of the condition and adjust treatment as needed.

The synthesis of the work underscores the shift towards a multidisciplinary approach, fostering interdisciplinary collaboration among pediatric ophthalmology specialists, pediatricians, medical geneticists, and other healthcare professionals to ensure comprehensive and tailored management for each case. By understanding the diversity of clinical presentations, healthcare professionals can more effectively anticipate individual needs of children and provide tailored care to optimize their visual health and quality of life.

Regarding therapeutic approaches, a rigorous evaluation of the various available options, including medical and surgical approaches, is necessary to determine the best strategy for each patient.

In the face of these challenges, sharing clinical experiences becomes essential to inform the management of these anomalies, with ongoing training necessary to ensure quality care. This discussion should be a dynamic forum where clinical

experience, cutting-edge research, and interdisciplinary collaboration converge toward optimal management, reflecting a commitment to medical excellence and informed practice.

4. Conclusion

The management of congenital optic disc anomalies in children requires an integrated and collaborative approach involving a diverse range of healthcare professionals. This multidisciplinary approach helps better understand the individual needs of patients and optimize their care. Sharing clinical experiences and integrating research advancements are essential to ensure quality care and improve patient outcomes. Additionally, ongoing training is necessary to maintain high standards of medical practice. By adopting an evidence-based approach and fostering collaboration among healthcare professionals, we can ensure better visual health and quality of life for children with congenital optic disc anomalies.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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