Case Report

Unilateral Optic Disc Pseudo-duplication with Contralateral Chorioretinal Coloboma: A Case Report

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Abstract

Optic disc pseudo-duplication (ODPD) is a rare condition presumed to be a form of retinal coloboma. In this report, we present the case of a child with ODPD in one eye and diffuse chorioretinal coloboma, along with microcornea in the fellow eye. This case supports the hypothesis that ODPD may be linked to the colobomatous nature of the condition.

This case highlights the unusual co-occurrence of optic disc pseudo-duplication and contralateral chorioretinal coloboma with microcornea in a young child. Multimodal imaging confirmed the congenital nature of both anomalies and excluded true optic nerve duplication. The findings support the growing hypothesis that optic disc pseudo-duplication may represent a variant of colobomatous malformation, likely resulting from incomplete closure of the embryonic fissure. Recognition of such associations is important for accurate diagnosis and genetic counseling.

Keywords: Optic Disc; Pseudo-Duplication; Coloboma; Case.

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Introduction

Optic disc duplication is a rare anomaly characterized by the presence of two separate apparent optic discs in one eye. However, most reported cases of optic disc duplication are not true duplications of the optic nerve and are referred to as pseudo-duplication. True optic nerve duplication is exceedingly rare in humans, with only a few reported cases. Nevertheless, multiple optic nerve fascicles have been observed in some primitive vertebrates, such as teleost fishes [1].

Reported cases of true optic disc duplication have been predominantly unilateral and associated with decreased vision in the affected eye [2]. Optic disc pseudo-duplication (ODPD) is typically a chorioretinal anomaly that resembles the optic disc and is located near the normal disc. Various congenital or acquired abnormalities can manifest as disc pseudo-duplication, including optic disc coloboma, peripapillary chorioretinal coloboma, inflammatory foci, chorioretinal atrophy, and scleral excavation in myopia [3,4].

In this report, we present a case of unilateral congenital ODPD associated with diffuse chorioretinal coloboma and microcornea in the contralateral eye. This unique association suggests that ODPD may represent a form of chorioretinal coloboma.

Case Report

A two-year-old girl was referred to our clinic for the evaluation of congenital epiphora in her left eye. She was the product of a non-consanguineous marriage, and her pregnancy had been normal, with term delivery and typical growth and development. During the first trimester of pregnancy, her mother had used amoxicillin and acetaminophen-codeine. The patient also had a history of urinary tract reflux disease.

Upon examination, her general appearance and mental status appeared normal, with the exception of large and anteriorly rotated external ears. In the external eye examination, the right eyelid fissure and the eye itself appeared smaller than the left side, and the patient was unable to fixate and follow with her right eye, while her left eye exhibited normal fixation and following abilities (Figure 1, A). Subsequently, she underwent an examination under anesthesia.

The full cycloplegic refraction revealed a refractive error of -6.00 in the right eye and +0.50 in the left eye. The horizontal corneal diameter was measured at 8 millimeters for the right eye and 12 millimeters for the left eye. Both eyes exhibited iris colobomas, with the right eye's coloboma being larger. Fundoscopy revealed a large chorioretinal coloboma involving the macula and optic disc in the right eye (Figure 1, B). In the left eye, a disc-like structure was observed inferiorly and slightly nasally to the actual optic disc. This structure appeared pale, was approximately one disc diameter in size, and was connected to the optic disc via the inferotemporal retinal vessels. Additionally, a large atrophic rim surrounded the pseudo-disc (Figure 1, C). Orbital and brain MRI scans were unremarkable, confirming the presence of a single optic nerve in each eye (Figure 1, D). The axial length of the right eye was measured at 21.2 mm, and the left eye at 20.9 mm. B-scan imaging demonstrated only one optic nerve stalk in the left eye (Figure 1, E). Examinations under anesthesia also revealed an imperforated Hasner membrane in the left nasolacrimal duct, which was successfully addressed through nasolacrimal duct probing.

A pediatric consultation was requested, and the patient underwent normal cardiac, endocrine,

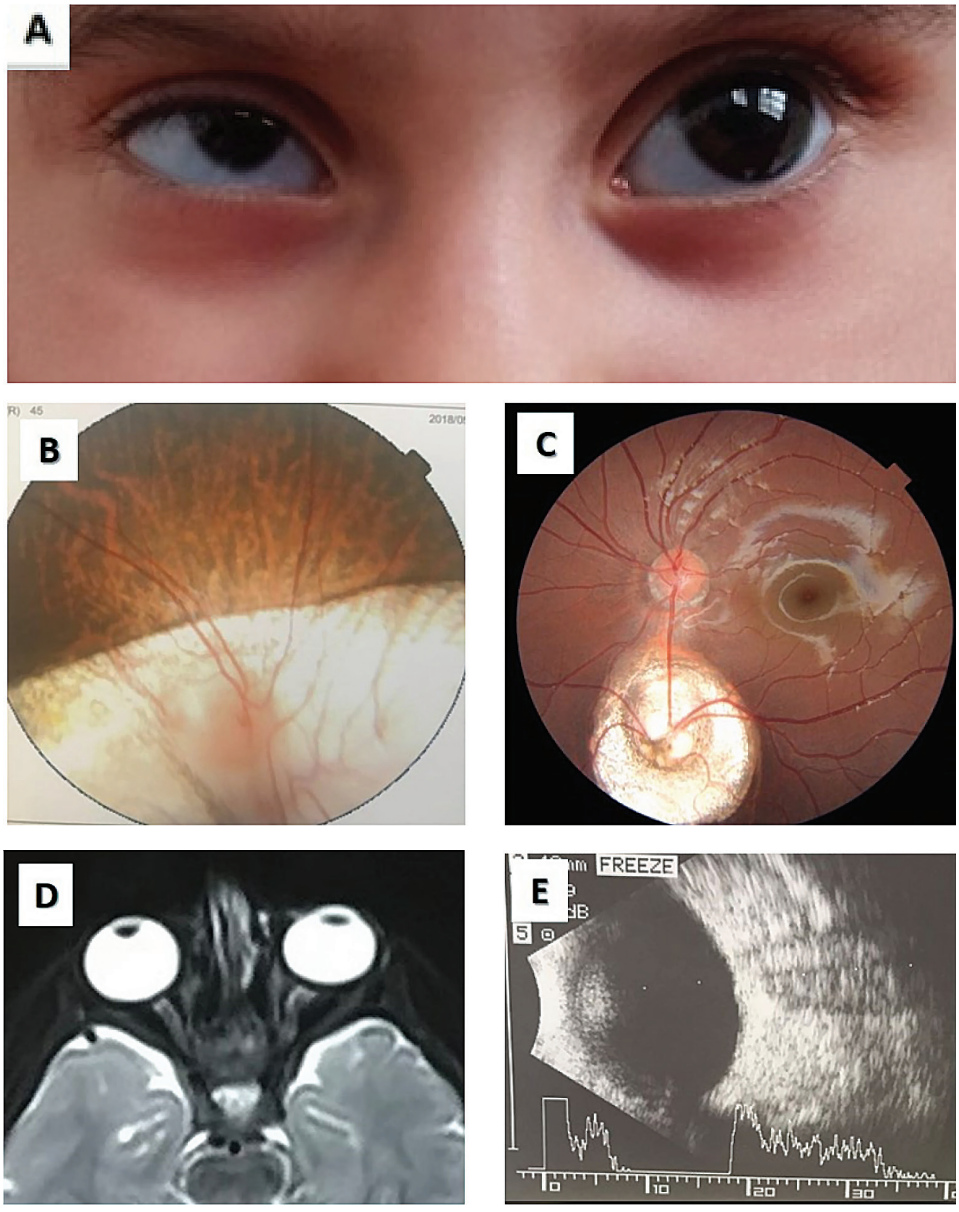

Figure 1 (A-E): In the external eye examination (A), the right eyelid fissure and the eye itself appeared smaller than the left side. Fundoscopy (B) revealed a large chorioretinal coloboma involving the macula and optic disc in the right eye. In the left eye (C), a disc-like structure was observed inferiorly and slightly nasally to the actual optic disc. Orbital and brain MRI scans (D) were unremarkable, confirming the presence of a single optic nerve in each eye. B-scan imaging (E) demonstrated only one optic nerve stalk in the left eye

neurologic, and hearing evaluations.

Discussion

In this report, we present a case of optic disc pseudo-duplication (ODPD) associated with contralateral large chorioretinal coloboma and micro-cornea.

Congenital ODPD is presumed to be a manifestation of a well-demarcated coloboma, typically located inferior to the normal optic disc and connected to it through bridging vessels [3]. Fluorescein angiography has demonstrated synchronous filling in these

vessels alongside the central retinal vessels [5,6]. On the other hand, acquired ODPD is described as chorioretinal atrophy and scleral excavation, often seen in cases of myopia. In these acquired cases, there is typically no bridging vessel connecting the actual and pseudo-discs [4,7,8]. The pathogenesis of acquired ODPD in myopia involves globe elongation leading to scleral stretching at the entrance of short posterior ciliary arteries, resulting in peripapillary lesions [8]. It has also been suggested that chorioretinal atrophy, scleral excavation, or defects in the ciliary artery entrance through the staphyloma contribute to the pathogenesis of acquired optic disc pseudo-duplication [7].

Orbital imaging techniques, such as ultrasound, CT scans, and MRI, can be instrumental in distinguishing true optic nerve duplication from ODPD by confirming the presence of two separate optic nerve stalks in the rare cases of true duplication. Additionally, in fluorescein angiography of the optic nerve head, the true optic disc typically shows late staining compared to the pseudo-disc [6]. In our patient, we conducted an MRI, which revealed one optic nerve on each side. To our knowledge, true optic nerve duplication has only been documented historically, and no recent cases have been reported in CT or MRI scans in humans. Islam et al., [2] have reported 11 cases of congenital ODPD and state that further investigations are unnecessary if bridging vessels are observed between the true and pseudo-optic discs [2].

In most reported cases of ODPD, the pseudo-disc is found inferior to the normal disc within the optic fissure territory, often accompanied by chorioretinal atrophy and pigmentary changes [5,6,9]. This specific location has led to the proposal that ODPD represents a well-demarcated chorioretinal coloboma involving the optic nerve head. This proximity enables vascular communication between disciform coloboma and the normal optic disc [9].

Similar to typical colobomas, the underlying pathogenesis appears to involve the failure of fetal fissure closure within the optic cup. Any disruption in the closure of the fetal fissure during early stages of pregnancy can potentially lead to bilateral chorioretinal coloboma. The mechanism behind unilateral or asymmetric cases remains poorly understood.

In our case, the presence of a substantial chorioretinal coloboma in one eye and ODPD in the other represents a unique finding. This observation suggests that coloboma may underlie the nature of ODPD, likely resulting from a failure in fetal fissure closure. A similar case involving bilateral optic disc coloboma in conjunction with unilateral ODPD has been previously reported by Mcloone and Buchanan [10]. Furthermore, the presence of a pseudo-disc situated above the normal disc in some cases may indicate an alternative hypothesis, such as inflammatory lesions in the peripapillary area, giving rise to acquired coloboma-like lesions [2].

While bilateral chorioretinal coloboma is commonly associated with microphthalmia, our patient exhibited normal axial lengths for her age in both eyes [11]. Microcornea is generally linked to microphthalmia but has rarely been reported in macrophthalmia cases [1,13]. The coexistence of coloboma with microcornea and normal axial length is referred to as anterior segment microphthalmos. In a large case series reported by Hornby et al.,[13] anterior segment microphthalmos was found to be at least as common as microphthalmos with coloboma.

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Notably, in our patient, the eye with the more

severe coloboma (the left eye) had a larger axial length compared to the right eye with ODPD, which is presumed to be another form of coloboma. This finding may suggest that the processes of fetal cup closure and axial globe growth are independent and unrelated to each other.

Conclusion

In conclusion, the coexistence of ODPD in one eye and chorioretinal coloboma with microcornea in the other eye, as observed in this case, represents a rare and unique association. This association lends support to the hypothesis that congenital ODPD may indeed be a form of colobomatous anomaly.

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Footnotes and Financial Disclosures

Conflict of interest:

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