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Case Report

Bilateral congenital macular coloboma: a case report

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Abstract: Bilateral congenital macular coloboma is a rare condition, with only a few cases documented in the literature. We report a case of a 62-year-old woman who presented to Quzhou People's Hospital in January 2022 with complaints of dryness and foreign body sensation in both eyes, along with a history of lifelong poor vision. Her best-corrected visual acuity was 20/500 in the right eye and 20/400 in the left eye, with a spherical equivalent refractive error of -9.0 diopters bilaterally. Anterior segment examination was unremarkable; however, fundus examination revealed well-demarcated chorioretinal atrophic lesions approximately four disc diameters in size at the macula of both eyes. Optical coherence tomography (OCT) demonstrated a large excavation involving the retinal and choroidal layers beneath the fovea, consistent with a diagnosis of bilateral congenital macular coloboma. The patient was treated with sodium hyaluronate eye drops for dry eye symptoms, while the coloboma itself required no intervention. In patients with unexplained visual impairment, thorough fundus examination and high-resolution OCT imaging are essential for accurate diagnosis.

Keywords: Congenital anomalies, bilateral, macular coloboma, poor vision, case report

Introduction

Ocular colobomas are congenital defects that may involve various ocular structures, including the iris, choroid, lens, retina, and optic nerve [1]. Macular coloboma refers to a well-circumscribed, oval or round atrophic lesion in the macular region, typically measuring 3-6 disc diameters in area [2, 3]. It is believed to result from the incomplete closure of the embryonic optic fissure, and intrauterine inflammation has also been suggested as a contributing factor. Congenital macular coloboma is rare, occurring in approximately 0.5-0.7 per 10,000 live births, and may be associated with other anomalies such as high myopia, strabismus, and hydrocephalus [2]. Bilateral involvement is especially uncommon. Here, we report a case of bilateral macular coloboma in a patient with high myopia and dry eye symptoms, highlighting the importance of comprehensive ocular examination in patients with unexplained visual impairment.

Case presentation

In January 2022, a 62-year-old woman presented to the ophthalmology outpatient clinic at Quzhou People's Hospital with complaints of dryness and foreign body sensation in both eyes over the past six months. She denied any other ocular symptoms but reported poor vision since childhood. Her family history revealed parental consanguinity; her parents were first cousins (their mothers were sisters) (**Figure 1**).

Her best-corrected visual acuity was 20/500 in the right eye (OD) and 20/400 in the left eye (OS). The spherical equivalent refractive error was -9.0 diopters bilaterally, and no improvement was achieved with optical correction. Axial lengths were 26.3 mm (OD) and 26.5 mm (OS), indicating high myopia. Slit-lamp examination revealed mild lid margin hyperemia, obstruction of the meibomian gland orifices, and mucous secretions. The anterior chamber was deep and clear, and the crystalline lenses were normal in both eyes (**Figure 2**).

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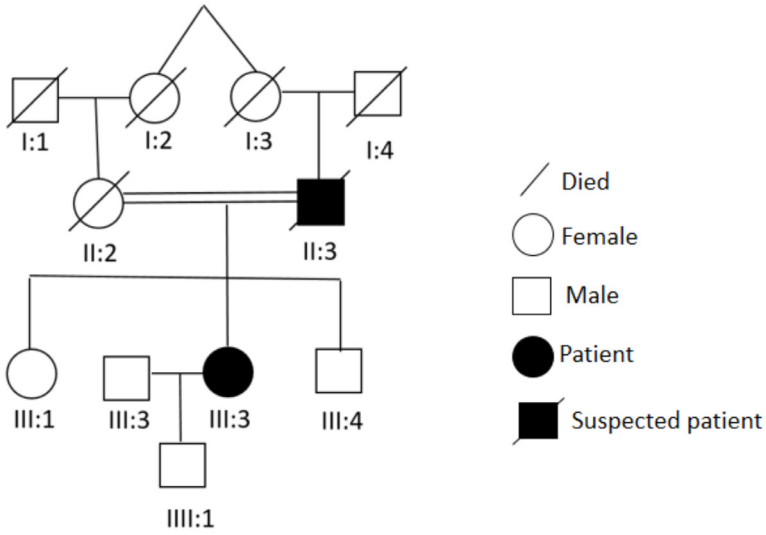

Figure 1. Pedigree of family.

Tear film break-up time was 4 seconds in the right eye and 5 seconds in the left eye (normal: ≥10 s). Schirmer's test showed tear secretion of 3 mm/5 min (OD) and 5 mm/5 min (OS) (normal: ≥5 mm/5 min), confirming the diagnosis of dry eye.

Fundus examination revealed well-defined pigmented chorioretinal atrophic lesions in the macular region of both eyes, measuring approximately four disc diameters and symmetrically located. The lesions were bordered by a dark, sharply demarcated rim of retinal pigment epithelium. The optic discs appeared normal, with typical color and cup-to-disc ratios. However, retinal vessels were slightly attenuated (**Figure 3**).

Optical coherence tomography (OCT) demonstrated large, crater-like foveal excavations with loss of the neurosensory retina and absence of the retinal pigment epithelium in the affected areas (**Figure 4A**, **4B**). Retinal nerve fiber layer analysis showed thinning in the inferotemporal quadrants of both eyes, with localized retinal nerve fiber layer thickness of 79 μm (OD) and 92 μm (OS) (**Figure 4C**, **4D**).

Serologic testing for Toxoplasma gondii antibodies was negative. The final diagnosis was bilateral congenital macular coloboma associated with high myopia and dry eye.

Currently, there is no effective treatment for macular coloboma. The patient was prescribed sodium hyaluronate eye drops and advised to apply warm compresses three times daily for dry eye management. At the 8-month follow-up, her visual acuity remained unchanged, and no progression of fundus lesions was noted.

Discussion

The present case underscores the importance of further investigation when a patient presents with poor visual acuity that cannot be adequately corrected. The etiology of congenital macular coloboma remains unclear. Two main hypotheses have been proposed: genetic predisposition and intrauterine infection. While most reported cases follow an autosomal dominant inheritance pattern, a few exhibit autosomal recessive transmission. Normally, macular differentiation occurs during fetal development; however, intrauterine infections-such as those caused by viruses, syphilis, tuberculosis, or Toxoplasma gondii - may disrupt the formation of arcuate nerve fiber bundles along the horizontal raphe, potentially resulting in macular coloboma [4, 5]. Since toxoplasmosis can produce chorioretinal scars resembling macular defects [6], anti-Toxoplasma antibody testing was conducted in this case, but yielded negative results. The patient's family history, which includes consanguineous marriage, suggests a genetic etiology. Unfortunately, her father's sample was unavailable, limiting further genetic testing and precluding genetic counseling for the family.

Based on the degree of scleral exposure and pigmentation within the lesion, Mann [7] classified macular coloboma into three types. In this patient, diffuse pigment hyperplasia was observed at the lesion margins without any associated choroidal or retinal vascular abnormalities, consistent with pigmented congenital macular coloboma.

Traditionally, diagnosis has relied on characteristic fundus findings, which are usually sufficient for clinical identification. However, funduscopy only provides a gross assessment of

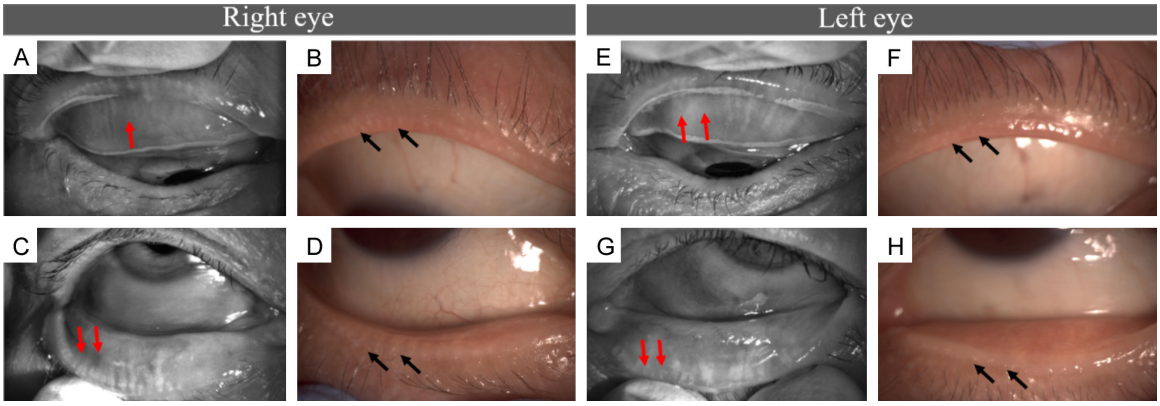

Figure 2. Infrared examination and anterior segmental photomicrographs. A-D. Right eye. E-H. Left eye. A, C, E, G. Infrared examination of the upper and lower eyelids reveals marked atrophy of the lid glands. B, D, F, H. Anterior segmental photomicrographs show mild congestion of the lid margins, obstruction of the lid gland openings, and sticky secretions. The red arrow indicates atrophy of the lid gland, while the black arrow points to a blocked lid gland opening with thick secretions.

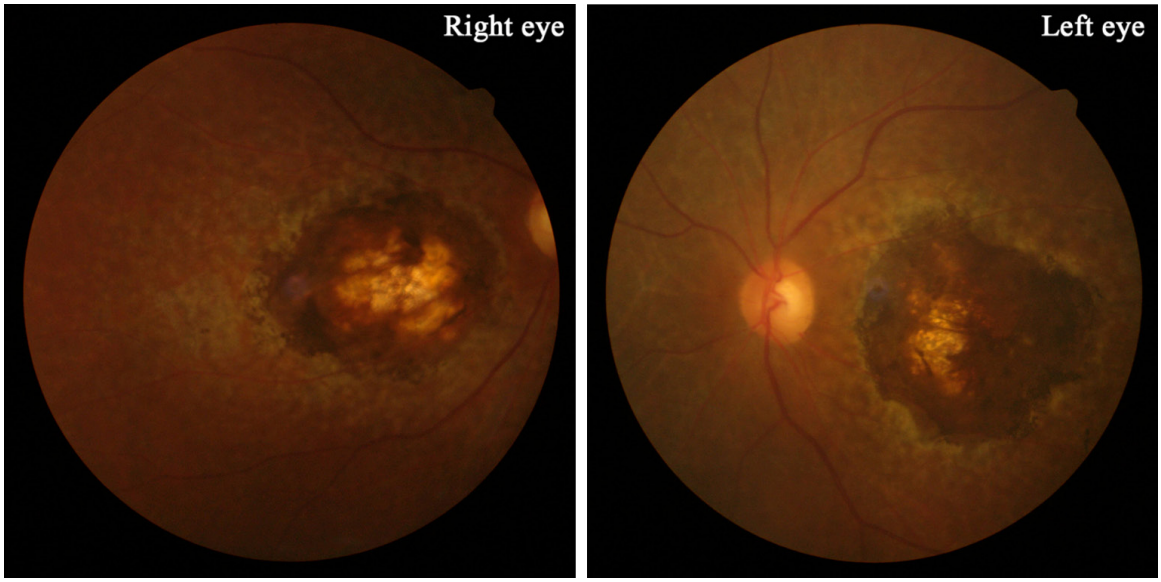

Figure 3. Fundus photographs of the patient's eyes.

the defect's size, extent, and presence of retinal detachment, and cannot accurately depict changes in the retinal architecture such as thickness or structural cleavage [3]. Optical coherence tomography (OCT), as a non-invasive and high-resolution imaging modality, offers a more detailed and intuitive approach to diagnosing and differentiating congenital macular coloboma [3].

OCT features of congenital macular defects typically include: posterior scleral ectasia; attenuation or absence of the capillary light band in the choroid at the lesion site; thinning or loss of the neurosensory retina, occasionally with inner and outer layer separation and interposed hyporeflective bands; close adherence of the neuroepithelium and underlying tissue at lesion margins; and glaucomatous-like cupping of the optic disc in cases where the coloboma involves the optic nerve head [8, 9].

In this case, OCT revealed a substantial interruption and absence of both the retinal and choroidal layers in the macular region, consistent with previous descriptions of pigmented

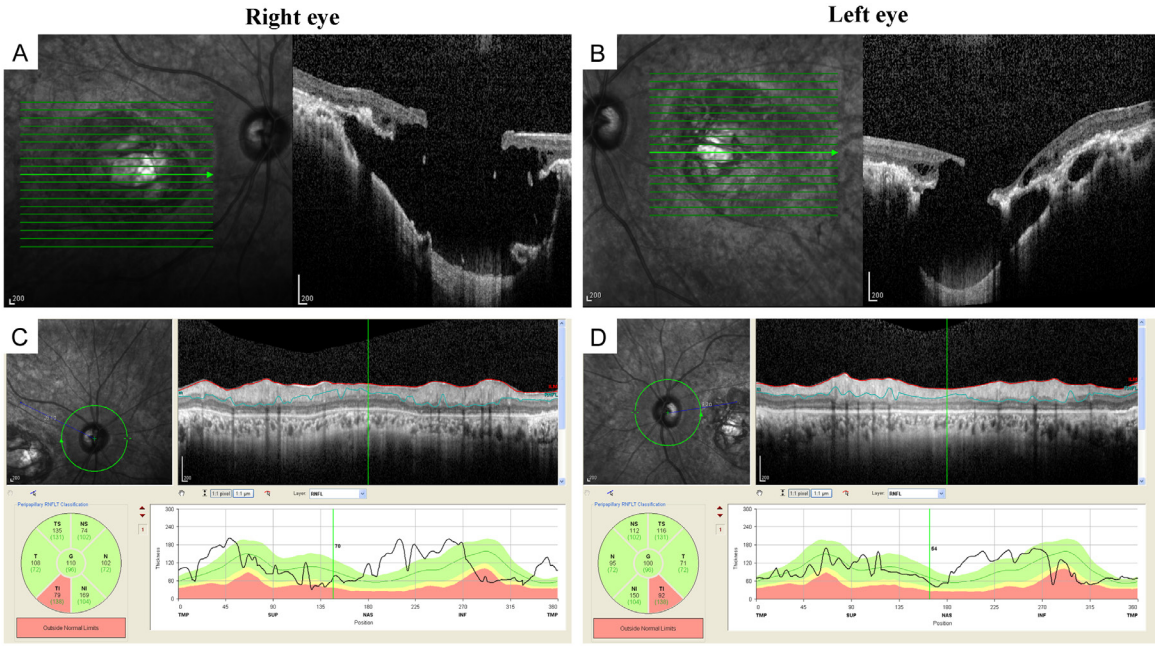

Figure 4. Optical coherence tomography. A, B. Macular lesions were visualized. C, D. The peripapillary retinal nerve fiber layer was observed in both eyes. S: superior; T: temporal; N: nasal; I: inferior.

macular coloboma. Abe et al. [9] reported similar OCT findings in a 4-year-old boy, showing complete loss of the retina and choroid, which aligns well with previous studies [10-13].

The patient had markedly reduced visual acuity and high myopia, with minimal improvement after refractive correction. A normal anterior chamber examination further indicated that the visual impairment stemmed from deeper retinal structural abnormalities. OCT imaging revealed a large, crater-like excavation at the fovea with total absence of the retina and choroid, explaining the poor visual outcome. This lesion could have been identified earlier had OCT been performed sooner; however, OCT is not routinely conducted in many optometry clinics. Therefore, patients with unexplained visual impairment, especially when anterior segment examinations are unremarkable, should undergo or be referred for OCT to avoid delayed diagnosis. Although early diagnosis may not significantly alter the prognosis-given the lack of effective treatment for macular coloboma - OCT remains the most reliable and efficient tool for detecting this condition.

Conclusion

Although no effective treatment currently exists for macular coloboma, our findings emphasize the importance of comprehensive evaluation in patients with uncorrectable bilateral visual impairment. In such cases, routine use of OCT should be strongly considered for early detection of macular structural abnormalities and to support timely diagnosis and monitoring, potentially delaying the progression of secondary symptoms.

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Disclosure of conflict of interest

None.

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