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Unilateral Rafflesia Arnoldii shaped Macular Coloboma Diagnosed with Optical Coherence Tomography: A Case Report

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ABSTRACT

Introduction: Macular colobomas are solitary, well-circumscribed, punched-out, crater-like lesions. They are usually bilateral and show various systemic associations. Case presentation: A 27-year-old female presented with painless diminution of vision in her left eye since birth without any family history of this condition. On Ophthalmological examination, the best corrected visual acuity (BCVA) was 20/20 and 20/200 in the right and left eye respectively. A well-defined, round, crater-like defect was seen in the macula of the left eye, with an unremarkable right eye. The optical coherence tomogram (OCT) showed a deep crater with absent retinal pigment epithelium and choroid in this region. Conclusion: The macular scars, such as toxoplasmosis or retinal dystrophies, can mimic this condition, and OCT can differentiate it with a few typical features.

KEYWORDS: *Macular scar, optical coherence tomography, unilateral macular coloboma*

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1. Introduction

Congenital macular coloboma is a rare condition that leads to a non-progressive diminution of visual acuity, affecting approximately 0.5-0.7/10,000 live births.[1] It is frequently found in the inferonasal quadrant, in line with embryological development, and is thus termed a typical coloboma.[2] It is a rare, congenital entity characterised by a round or oval area of chorioretinal atrophy of variable size at the macula. [3] In literature, various systemic associations of macular coloboma have been reported, such as Down's syndrome [4]..

2. Case presentation

A 27-year-old female presented with painless diminution of vision in her left eye since birth. On ophthalmological examination, the best corrected visual acuity (BCVA) was 20/20 and 20/200 in the right and left eye, with refractive error of -0.50 cyl and 1.50sph/-0.75cyl, respectively. According to the Hirschberg test, an exotropia of 20 degrees was observed in the left eye. There was a grade three relative afferent pupillary defect in the left eye. The rest of the anterior segment examination was unremarkable in both eyes. On fundus examination, a well-defined, round, crater-like defect was seen in the macula of the left eye, which was illuminated, yellow-ish, coarsely pigmented and atrophic. This was seen as a flower petal-like pattern arranged in a circle, measuring approximately one and a half optic disc diameters in height and width. At the centre of the excavated area, large choroidal vessels with bare sclera at the base were seen dis-turbing the retinal vasculature with enhanced backscattering. The entire architecture mimics Rafflesia arnoldii, the world's largest parasitic flower, more commonly known as the stinking corpse lily. Rafflesia arnoldii spreads underground as fine roots, which invade their host plants' roots. This takes water and nutrients from its host, and the cabbage-shaped buds emerge from the ground and take 21 months to grow completely, and then the flower finally emerges. It lasts for only one week.[5] [Figure 1 A]. Spectral-domain optical coherence tomogram (OCT) revealed the absence of retinal pigment epithelium (RPE) and choroid in the macular region, with disrup-tion of the retinochoroidal complex and atrophic neurosensory retina in the left eye. Enhanced backscattering was also noticed due to bare sclera and choroidal vessels [6] [Figure 1B]. The pa-tient is being followed up to rule out its association with cataracts, glaucoma, or any other comorbidities.

3. Discussion:

Macular colobomata are a consequence of incomplete differentiation of the arcuate bundles along the horizontal raphe. [2] They are atypical colobomas as they do not belong to the areas of embryon-ic cleft. The coexistence of macular coloboma with other retinochoroidal disorders, such as ret-initis pigmentosa, Leber amaurosis, and retinal degeneration, has been reported. [7] There have been reports linking macular coloboma to Toxoplasma and Zika virus infections.[8] The OCT performs cross-sectional imaging at a resolution of 10 to 20 μm of biological tissue. In this case, OCT showed an absent choroid and RPE and atrophic neurosensory retina, which is similar to a histological section and thus can be beneficial for the diagnosis of macular coloboma without a histologic section. [9]

4. Conclusions:

Unilateral congenital macular coloboma is a rare condition that presents with poor visual function and carries a very poor visual prognosis. This case is unique because it was observed in a young female and exhibited a distinctive pattern of Rafflesia flowers on the fundus, as confirmed by OCT showing the typical histological sections with absent retina and choroid. These patients should be managed with low vision aids and should be followed up to detect any other ocular morbidities eventually.

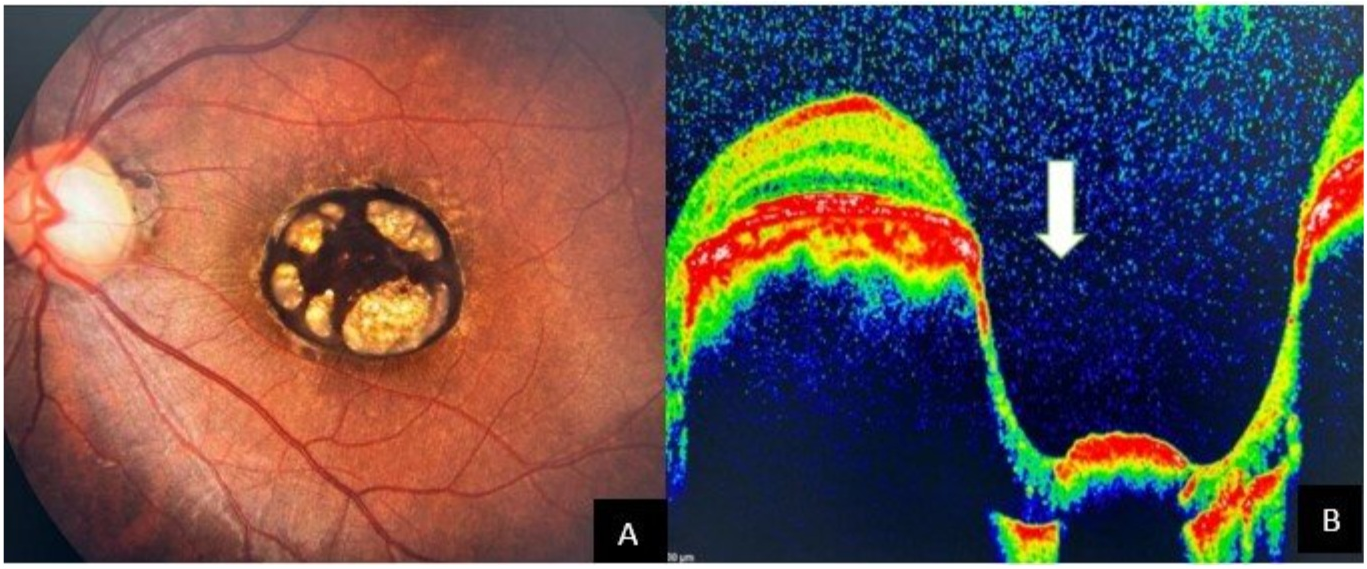

Figure 1A: Fundus examination showing macular coloboma as a solitary, punched-out, flower petals-like pattern arranged in a circle

(Rafflesia-like). B: Optical coherence tomography depicting the disruption of the retinochoroidal complex with the absence of retinal pigment epithelium and choroid in the macular region.

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