

Triple Fundal Coloboma: irregular closure of the embryonic cleft in the optic cup

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DESCRIPTION

A 45-year-old woman on routine evaluation was found to have coloboma of optic nerve head and two inferior choroidal colobomas (CCs) connected by a bridging retinal tissue in the left eye (figure 1A). Vertical optical coherence tomography (OCT) line scan through the CCs revealed thin rudimentary retinal tissue typical of an intercalary membrane along with an absent choroidal tissue and ectatic sclera. Notably, a 'hump' was seen between the CCs corresponding to the bridging tissue (figure 1B). Horizontal scan through the 'hump' showed a poorly developed thin retinal tissue with indecipherable layers and an underlying maldeveloped choroid with patchy presence of choroidal vessels (figure 1C). The fovea was normal. Right eye fundus was normal. Best-corrected visual acuity and anterior segments were normal in both the eyes.

Normal closure of the embryonic cleft in the optic cup occurs inside out and in anterior to posterior direction.¹ Some authors have stated that the closure begins in the ciliary body area and proceeds both ways along its long axis.¹ Defective closure can result in CC, optic nerve head coloboma and persistence of fetal vasculature.² The presence of multiple colobomas and partially developed chorioretinal tissue in our case are supportive of the postulated patchy closure of the fissure.² OCT of a normal appearing bridging tissue, revealed partial closure with chorioretinal remnants. Similar OCT features of CC have been seen previously, though the bridging tissue lacked detailed evaluation.³

Therefore, OCT imaging of multiple colobomas and the intervening transition zone presents a rare

but very good opportunity to better understand closure of the embryonic optic cup cleft.

Learning points

- ▶ Defective and interrupted closure of the embryonic cleft in the optic cup may lead to multiple colobomas.
- ▶ Optical coherence tomography of the clinically normal looking bridging retina may reveal an incomplete closure as well.

Contributors RB was involved in diagnosis, imaging and review with approval of final draft of manuscript. BT was involved in interpretation of images and review with approval of final draft of manuscript. SVA was involved in interpretation of images, writing and review with final approval of final draft of manuscript. RC contributed to interpretation of images, writing and review with approval of final draft of manuscript. SVA is responsible for the overall content as guarantor.

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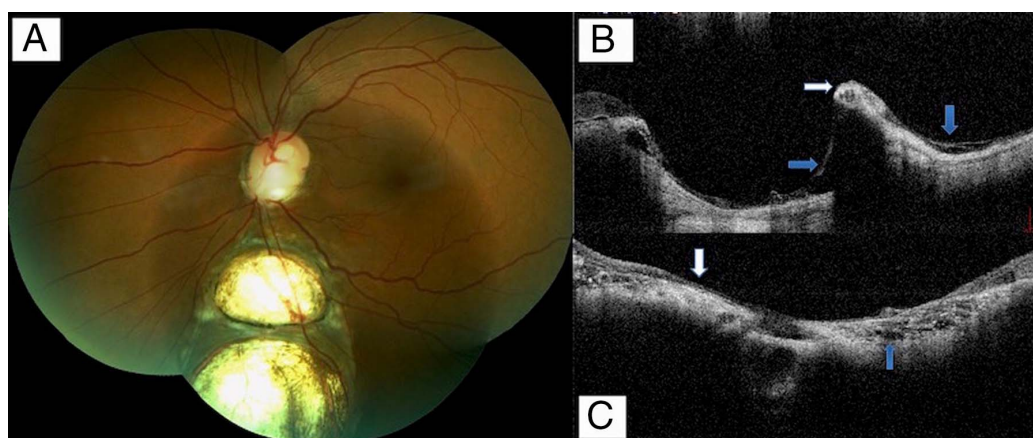


Figure 1 (A) Triple fundal coloboma: optic nerve head coloboma with two choroidal colobomas. Characteristic choroidal atrophy can be seen surrounding the colobomatous area in the optic nerve head. (B) Thin rudimentary retinal layers typical of an intercalary membrane (blue arrow) with hump (white arrow) between the two colobomas. (C) Bridging tissue showing peripheral retinal (white arrow) and choroidal remnants (blue arrow) in a shallow ectatic sclera.



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