A 22-Year-Old Male with History of Nystagmus, Ocular Albinism, and Foveal Hypoplasia

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Introduction:

A 22-year-old white male presented to the eye clinic for a routine exam. His past ocular history was significant for nystagmus, ocular albinism, and foveal hypoplasia. He had no new complaints. His exam remained stable: 20/60 vision in both eyes with unchanged horizontal nystagmus, transillumination defects of both irides, and blonde fundi without associated retinal tears or detachments. Optical coherence tomography (OCT) revealed foveal hypoplasia of both maculae with otherwise normal retinal contour and thickness (Figure 1). Curiously, Optos widefield images obtained as part of routine funduscopic screening revealed bulls-eye patterned waves within each macula (Figure 2). These were unchanged over the course of two annual visits and were not visible on funduscopic examination.

Discussion:

This pattern has been described in recent literature as the "concentric macular rings" sign.[1,2] It is thought to derive from the more-upright alignment of the outer plexiform layer within the macula (also known as nerve fiber layer of Henle) in patients with foveal hypoplasia when compared to those with normal foveae. During foveal development, inner retinal layers migrate away from the fovea while cone nuclei pack closely within the outer nuclear layer.[3] These migrations lead to elongation and diagonality of cone axons extending to the dendrites of bipolar and horizontal cells. In patients with even low-stage foveal hypoplasia, this migration is disrupted which leads to vertical cone axons.[4] This misalignment is speculated to confer properties of polarity and birefringence of the fiber layer which can then be detected by Optos imaging which scans with polarized light.[2]

Interestingly, this sign appears to be exceptionally sensitive and specific to foveal hypoplasia. In one study of fifty-five eyes with foveal hypoplasia of various etiologies, it was detected in 100% of eyes with foveal hypoplasia and undetectable in eyes that were either normal or with any other macular pathology.[2] This was true even in instances of unilateral foveal hypoplasia. This suggests that Optos widefield photography could be a useful diagnostic tool for patients without the diagnosis of foveal hypoplasia but with unexplained poor vision where nystagmus prohibits proper fixation on an OCT target.

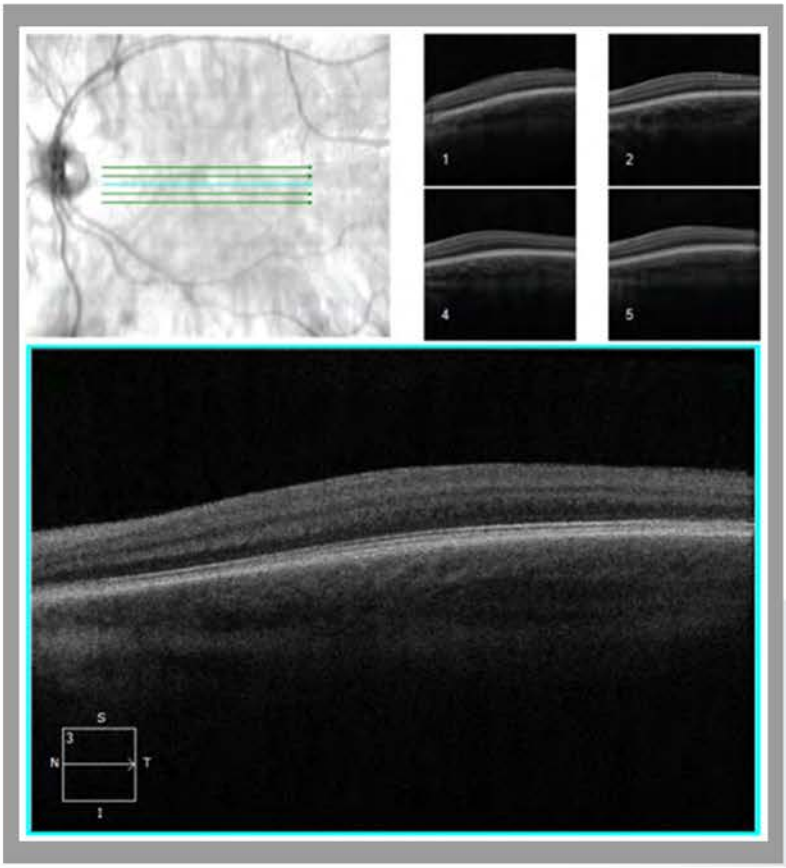
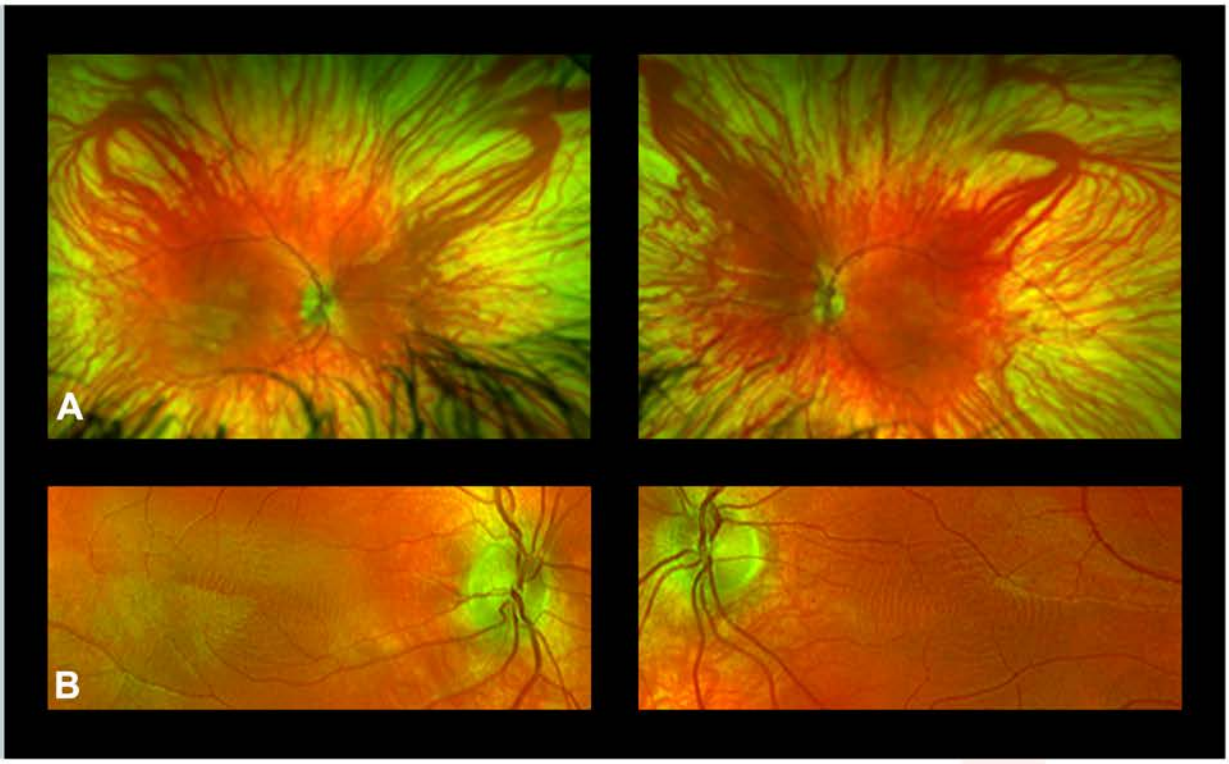


Figure 1: Five-line raster scan of the left eye obtained on Cirrus OCT demonstrates absence of the fovea but otherwise normal retinal layers. The right eye was symmetric in appearance.

Figure 2A): Optos wide-field images demonstrate blonde fundi without other retinal pathology.

Figure 2B): Enhancement on the foveae demonstrates absence of foveal pit and concentric rings within the macula of both eyes.



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References:

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