A 35-Year-Old Male with an Abnormal Foveal Contour

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Introduction:

A 35-year-old male was referred for evaluation of abnormal foveal contour.

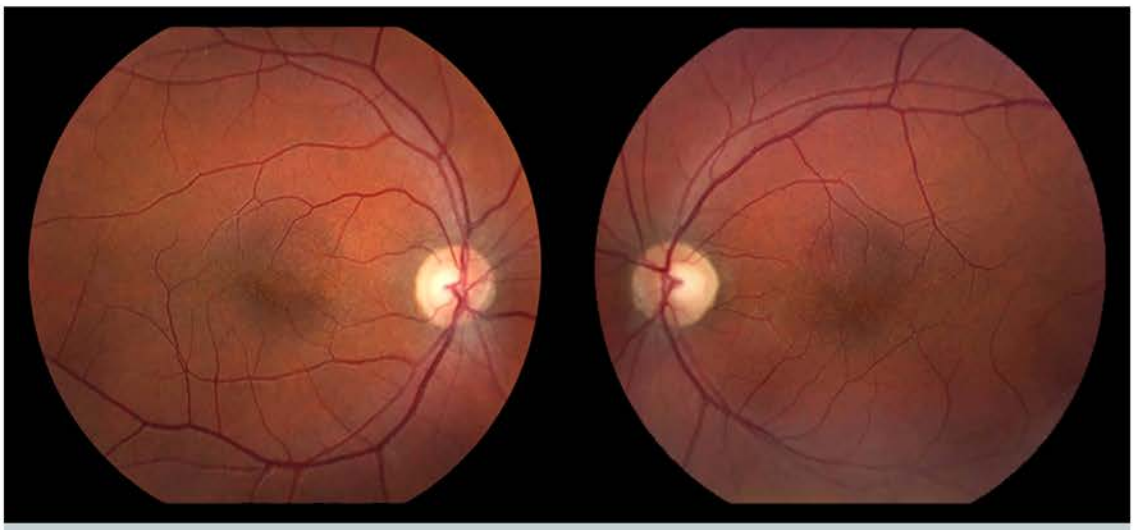

Figure A: Color photos of the posterior pole of each eye revealing an overall normal appearance.

Exam:

Best corrected visual acuity was 20/25 in the right eye and 20/30 in the left eye. Intraocular pressures, visual fields, and extraocular motilities were normal. No afferent pupillary defect was present. Anterior segment exam was normal. Dilated fundus examination revealed an overall normal appearing posterior pole but lacking a foveal light reflex (Figure A). Fundus autofluorescence revealed mild mottling in the left eye (Figure B). Optical coherence tomography (OCT) and optical coherence tomography angiography (OCTA) revealed the most striking finding of the absence of a foveal pit and foveal avascular zone bilaterally (Figure C & D).

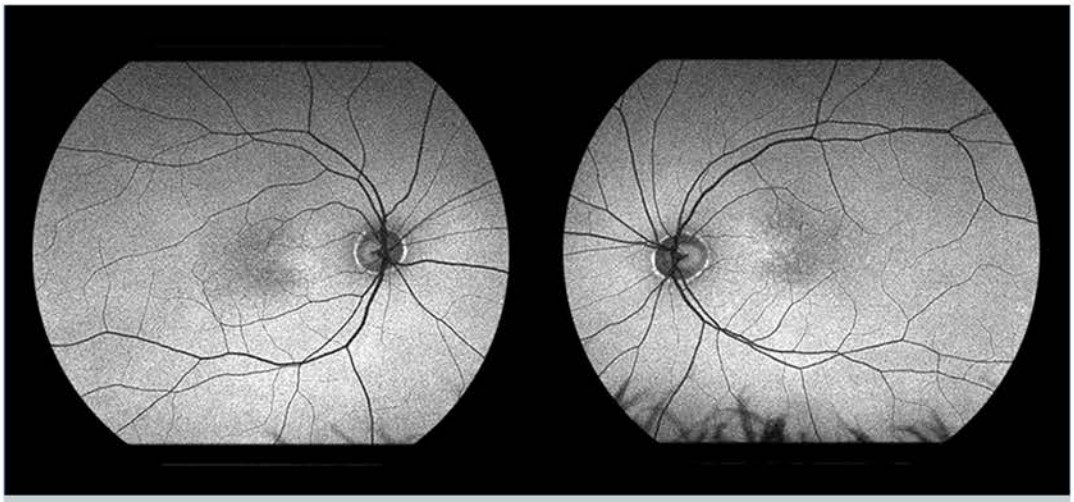

Figure B: Fundus autofluorescence of each eye revealing normal auto fluorescence of the right eye and mild mottling in the left eye.

Discussion:

This patient was diagnosed with fovea plana. The differential diagnosis of these findings includes that of foveal hypoplasia which can occur in ocular albinism,[1] aniridia,[2] nanophthalmos,[3] incontinentia pigmenti,[4] ROP,[5] Goldenhar-Gorlin

syndrome,[6] as well as more rare genetic disorders.[6] Historically, this condition has had significant overlap with foveal hypoplasia which can be associated with significant visual impairment along with characteristic signs of each of the aforementioned conditions. A blond fundus, iris transillumination defects, nystagmus, and photophobia may be seen in ocular albinism.[1] Aniridia may show a hypoplastic or absent iris depending on the severity.[2] Hyperopia secondary to decreased axial length, microcornea, shallow anterior chambers, and crowded optic nerves may be seen in nanophthalmos.[3] Incontinentia pigmenti can present with skin lesions, teeth defects, peripheral retinal avascularity, and macular occlusive disease in female patients.[7] Findings seen in patients with a history of ROP include dragging of the macula and vasculature, retinal pigmentary changes, lattice-like degeneration, myopia, amblyopia, and tractional retinal detachments.[8] In Goldenhar-Gorlin Syndrome, the patient may exhibit microphthalmia, limbal dermoids, and preauricular tags or pits.[9]

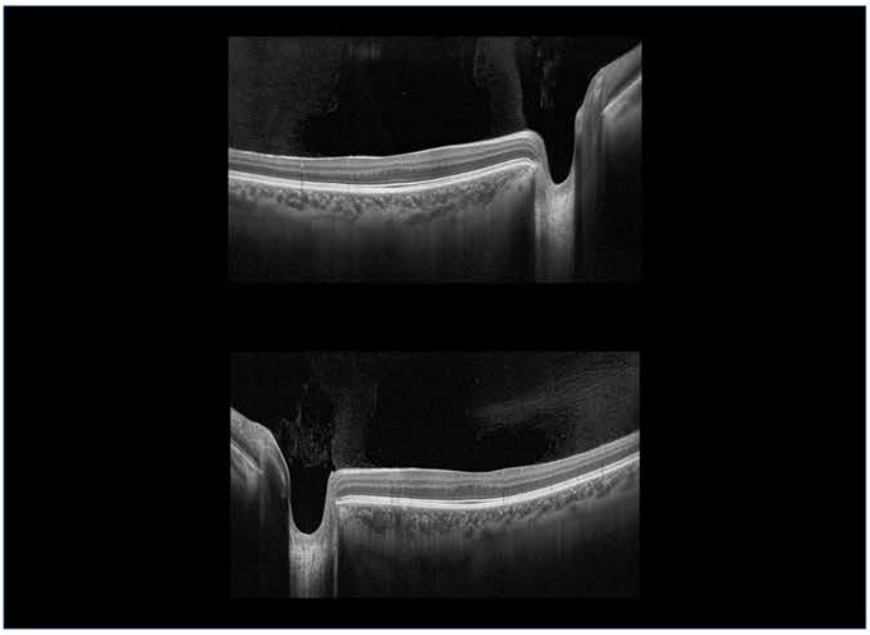

Figure C: OCT of each eye. Notice the lack of foveal depression. The foveal ellipsoid zone is intact. There is retinal thickening in the area of the fovea secondary to more prominent inner retinal layers and thickening of the outer plexiform layer.

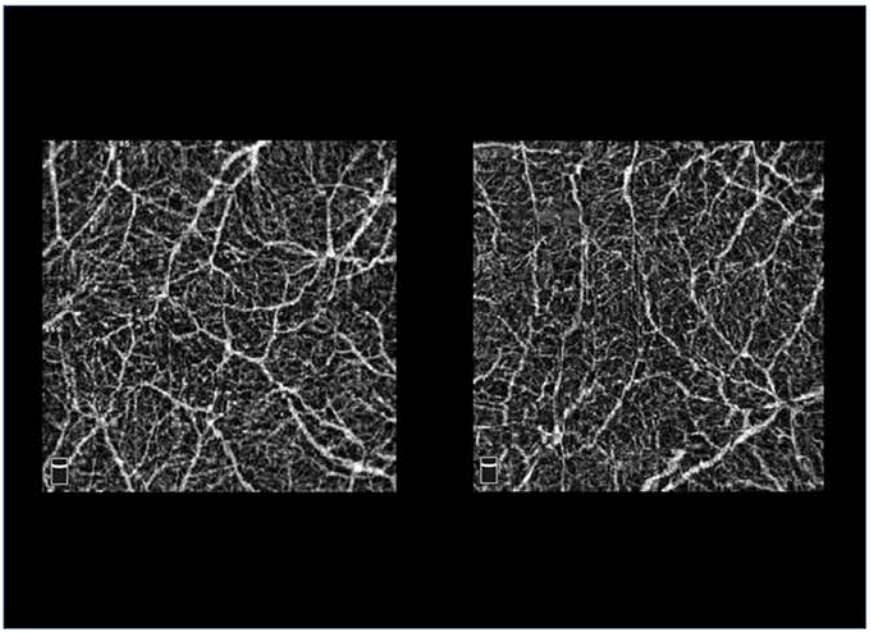

Figure D: Optical coherence tomography angiography of the fovea in both eyes revealing the lack of a foveal avascular zone.

Recent literature by Marmor et al has attempted to separate the two entities as visual outcomes of foveal plana are typically good compared to the variable but significant impairment that can be associated with foveal hypoplasia.[2] Patients with fovea plana typically have a visual acuity ranging from 20/20 to 20/50, and their examination is notable for the absence of a foveal light reflex on exam. OCT imaging is quite useful as it reveals the absence of the foveal pit. This lack of depression is due to the inner retinal layers are not thinning in the foveal region, thickening of the outer nuclear layer. Elongation of the photoreceptor zone may also be seen. When viewed with fluorescein angiography or optical coherence tomography angiography, a lack of foveal avascular zone is apparent.[2] Electrophysiological studies (mfERG) reveal normal waveforms across the posterior pole.

Combined, these findings reveal that although not anatomically normal, these eyes are physiologically

capable of fine acuity vision.[2] Still though, the appropriate workup should be undertaken to rule out causes of foveal hypoplasia especially if this finding is diagnosed in a pediatric patient.

References:

1. Dorey S, Neveu M, Burton L, Sloper J, Holder G. The clinical features of albinism and their correlation with visual evoked potentials. Br J Ophthalmol. 2003;87(6):767-772.

2. Marmor M, Choi S, Zwadzki R, Werner J. Insignificance of Foveal Pit in Humans. Arch Ophthalmol. 2008;126(7):907-913. doi:10.1016/j.biotechadv.2011.08.021.Secreted

3. Walsh M, Goldberg M. Abnormal foveal avascualr zone in nanophthalmos. Am Acad Ophthalmol. 2007;143(6):1067-1068.

4. Goldberg M, Custis P. Retinal and other manifestations of incontinentia pigmenti (Bloch-Sulzberger syndrome). Ophthalmology. 1993;100(11):1645-1654.

5. Recchia F, Recchia C. Foveal dysplasia evident by optical coherence tomography in patients with a history of retinopathy of prematurity. Retina. 2007;27(9):1221-1226.

6. Querques G, Pierro L, Zucchiatti I, Corbelli E, Querques L, Bandello F. Multimodal Imaging of Diabetic Retinopathy in a Patient with Fovea Plana. Retina. 2016;36(10):e93-e94. doi:10.1097/IAE.0000000000001020

7. Christian C, Swinney B, Han D, Karth P. Incontinentia Pigmenti: A Comprehensive Review and Update. Ophthalmic Surgery, Lasers, Imaging Retin. 2015;46(6):650-657.

8. Smith B, Tasman W. Retinopathy of prematurity:late complications in the baby boomer generation. Trans Am Ophthalmol Soc. 2005;103:225-236.

9. Bhallil S, Benatiya I, El Abdouni O, Mahjoubi B, Hicham T. Goldenhar syndrome: ocular features. Bull Soc belge Ophtalmol. 2010;316:17-20.

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