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Case Report

Unilateral Isolated Foveal Hypoplasia

Adam B. Jacobson[1], Thaddeus L. Wandel[1], and Alan D. Springer[1,2]*

[1]Department of Ophthalmology, New York Medical College, USA
[2]Department of Cell Biology & Anatomy, New York Medical College, USA

*Corresponding author
Alan D. Springer, Department of Cell Biology & Anatomy, New York Medical College, Valhalla, NY 10595, USA, Tel: 914-594-4037; Fax: 914-594-4653; Email: alan_springer@nymc.edu

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Abstract

Purpose: Foveal hypoplasia is described as an absent or shallow pit, thickened inner retina, shortened outer segments, and an increased retinal thickness. Bilateral foveal hypoplasia is often associated with other ocular comorbidities, including albinism, aniridia, microphthalmia, and retinopathy of prematurity. We are reporting a case of unilateral isolated foveal hypoplasiain an adult, using high resolution Cirrus OCT raster scans.

Observations: The scan showed a patient with an epiretinal membrane, no evidence of macular edema, and an absent foveal pit in one eye. The scan also revealed thick inner retinal laminae overlying a fovea externa with outer segments that had normal lengths, not normally seen in uncomplicated cases of epiretinal membranes.

Conclusions: A macular epiretinal membrane (ERM), rippling of the retina under the ERM, and an absent foveal pit are usually interpreted as being caused by traction and edema. Quantitative analysis of the OCT suggests that this patient should be diagnosed as having unilateral foveal hypoplasia that is independent of the ERM. High resolution scans (4,096 A-scans/B- scan) can help distinguish macular edema from foveal hypoplasia.

ABBREVIATIONS

OCT: Optical Coherence Tomography; ERM: Epiretinal Membrane; BCVA: Best Corrected Visual Acuity

INTRODUCTION

Foveal hypoplasia is a retinal morphology reflecting variably stunted macular development. The mal developed fovea may have a shallow or absent pit [1-7], thickened inner retinal layers [1-3,5-7], a thin outer nuclear layer [5,6], shortened outer segments [4,6] or an overall increased retinal thickness [2,3,5,6]. In addition, several congenital anomalies are often associated with foveal hypoplasia, including albinism [6,8-12], aniridia [10,13], microphthalmos [12], achromatopsia [6,14], and retinopathy of prematurity [15]. Cases of isolated foveal hypoplasia, without any of the three preceding comorbidities, have also been described [4,6,8,10,16,17]. To date, only two cases of isolated unilateral foveal hypoplasia have been reported, both in young individuals. These cases were also associated with poor vision and numerous anatomical abnormalities in the affected eyes [18,19].

Optical coherence tomography is effective for detecting, describing, and quantifying foveal hypoplasia [6,11]. Higher resolutions obtained with spectral domain OCT permit more detailed views of the retinal layers, compared to the previous generation of lower resolution time domain OCT [20].

CASE PRESENTATION

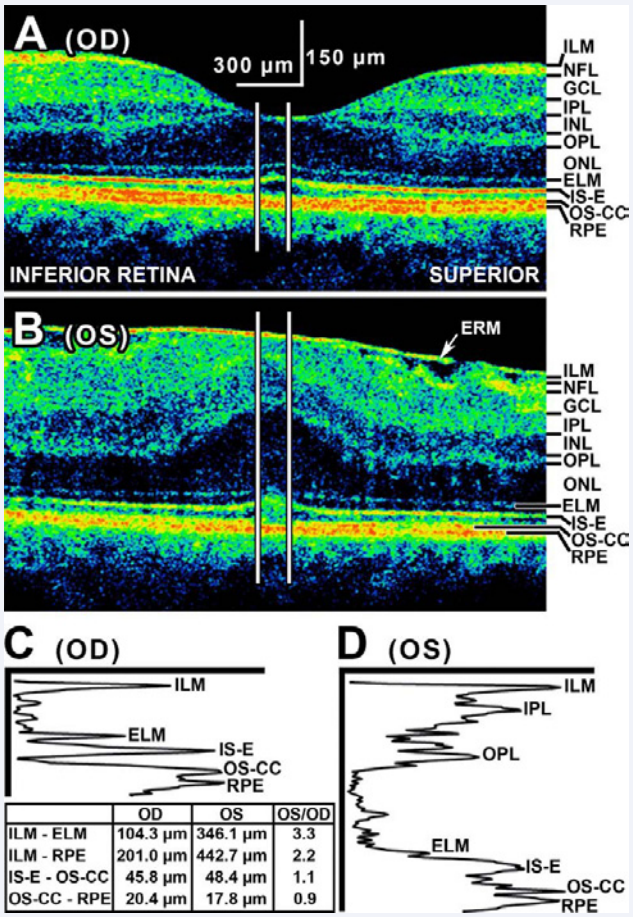
WD, a 68 year old female, was seen over three years. Her ophthalmological history consists of bilateral posterior vitreous detachments, glaucoma suspect, and uncomplicated cataract excision. Her clinical records show a stable corrected visual acuity (OU: 20/20), even with the ERM shown below. Over three years, her BCVA in the affected eye diminished to 20/400.

Optical coherence tomography was obtained over the course of her clinical observation. The first OCT, several years prior to cataract surgery and ERM stripping, showed unilateral foveal hypoplasia and an ERM overlying the hypoplastic fovea. Scans of her right eye showed normal foveal morphology.

Only the first OCT scan had an extremely high resolution. Vertical, Cirrus OCT HD 5 line raster scans that were 6 mm long and with each line-scan comprising 4,096 A-scans are shown in Figure 1. The right fovea showed a normal pit (Figure 1A). However, the left fovea was dome shaped (Figure 1B), contained all of the inner retinal laminae, and lacked a pit.

Axially-oriented pixel intensity scans, centered on the fovea (Figure 1C, D), allowed quantification of laminar thicknesses and distances between laminae for each retina (Figure 1C, D). The hypoplastic left fovea was 3.3 times thicker than the right, as measured from the inner to the external limiting membrane

(Figure 1C, D). ONL thickness (distance between outer edge of OPL and ELM) for the left eye was 183.2 µm and104.3 µm for the right eye, a 1.8-fold difference. By contrast, photoreceptor inner and outer segment lengths for each fovea were comparable (Figure 1C). Corrected acuity for both eyes was 20/20 when the scans were performed. Nevertheless, when asked whether she perceived any functional difference between her eyes, the patient volunteered that the acuity in her left eye had been poor since childhood.



	OD	OS	OS/OD
ILM - ELM	104.3 µm	346.1 µm	3.3
ILM - RPE	201.0 µm	442.7 µm	2.2
IS-E - OS-CC	45.8 µm	48.4 µm	1.1
OS-CC - RPE	20.4 µm	17.8 µm	0.9

Figure 1 Central 2.5 mm portions of 6 mm long vertical Cirrus 5 line HD raster scans. **(A)** OD has a normal foveal morphology. **(B)** OS foveal region has an ERM, lacks a pit and contains a continuous band of inner retinal laminae. Outer retinal laminar labelling after Spaide & Curcio (24). OCT signal strength for A &B is 9 & 8 respectively. **(C & D)** Longitudinal reflectance profile of a 102 µm wide vertical band centered on the fovea of scans converted to grayscale and rotated so that the ILM was horizontal to correct for tilt at the fovea. The approximate locations of the scans aremarked with vertical lines in **A** and **B**. Major peaks in the profiles are labeled and distance between some of the peaks are presented in C. All of the laminae visible parafoveally are also evident in the OS "fovea".

Abbreviations: ILM: Inner Limiting Membrane; NFL: Nerve Fiber Layer; GCL: Ganglion Cell Layer; IPL: Inner Plexiform Layer; INL: Inner Nuclear Layer; OPL: Outer Plexiform Layer; ONL: Outer Nuclear Layer; ELM: External Limiting Membrane; IS-E: Inner Segment Ellipsoid Region; OS-CC: Outer Segment Contact Cylinder Region; RPE: Retinal Pigment Epithelium; OD: Right Eye; OS: Left Eye; ERM: Epiretinal Membrane

The ERM (Figure 1B) produced rippling of the inner retinal parafoveal region. The OCT scans did not show signs of retinal detachments or hyporeflective, fluid filled spaces, indicative of edema. There was also no indication of a pit that was tented inwardly. Furthermore, mean pixel intensity across the ONL for both eyes was similar (Figure 2A, B).

WD was subsequently lost to follow up, and additional images were not acquired.

DISCUSSION

We are reporting the first case of unilateral foveal hypoplasia in an adult. There are four observations of WD's left eye that led to this diagnosis. First, the obvious lack of a foveal pit. Second, the left eye had a thickened outer nuclear layer at the fovea. Third, the overall retinal thickness of the left eye was increased 3.3-fold compared to the unaffected eye. Fourth, and most notably, is the persistence of thick inner retinal layers at the fovea – a pathognomonic feature of foveal hypoplasia. Asakawa and Ishikawa [18] have recently presented another case of unilateral foveal hypoplasia in an infant with 20/100 vision in the affected eye; however, their hypoplastic macula does not show a fovea externa, and therefore we cannot be certain that the image is centered over the fovea.

These foveal features are consistent with previous descriptions of hypoplasia [1-7]. However, the possibility that these scans actually showed macular edema secondary to the ERM, must be considered. OCT findings of the macula include the absence of a pit and separation of outer nuclear layer from the outer plexiform layer by a hyporeflective collection of fluid [21,22]. To rule out the presence of edema, average pixel intensity of the outer nuclear layers in the HD OCT images was measured (Figure 2). Average pixel intensity should be lower, if edematous fluid was present. However, ONL pixel intensities of each eye were similar to one another. This suggests that the left retina was not deformed by macular edema secondary to traction by the ERM.

An alternative explanation to unilateral foveal hypoplasia is that the OCT scan of the left eye was not centered on the fovea. We consider this possibility unlikely since a fovea externa [23,24], an indicator of the center of the foveola, is present in both OCT scans (Figure 1A and B). Furthermore, IS-E to OS-CC distances, a measure of outer segment length, were comparable for both foveas (Figure 1C table). Moreover, remaining 4 line scans, nasal and temporal to those shown in Figure 2A, 2B had thinner ONLs and all lacked a fovea externa.

This case raises the question of whether unilateral foveal hypoplasia is rare or that its detection is related to OCT scanning resolution. Current OCTs have several scanning protocols that vary in resolution. For example, more retinal detail is resolved as the Cirrus protocol is changed from 200x200 cube scan to 512x128 cube scan to HD 5 line raster scan. We believe that in most cases, a unilateral ERM suggests a diagnosis of edema-induced foveal deformation. When an ERM deforms a normal fovea, the flattened floor of the pit is elevated and separated from the underlying outer retina, thereby leading to edema. However, in our case, there was no evidence of a previously existing pit, or of a retinal separation or hole. Furthermore, we used a 5 line HD raster scan instead of a lower resolution cube scan, which permitted visualization of the un displaced inner retinal laminae.

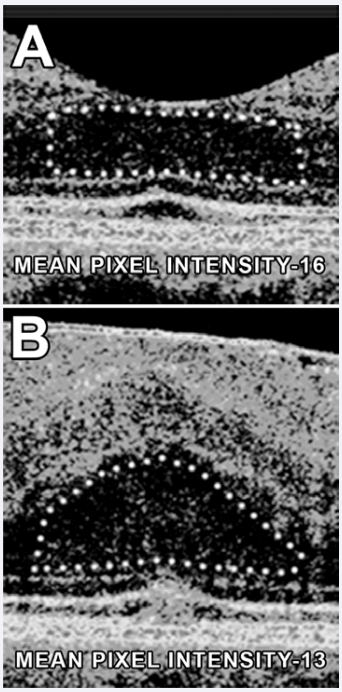


Figure 2 Foveal regions from Figure 1A, B converted to gray scale. Mean pixel intensity in the outer retinal area bounded by the dotted lines was calculated and is at the bottom of each figure. The values were similar, providing no evidence for a fluid filled/edematous area in the hypoplastic retina.

Had a lower resolution scan been done, we might have decided that edema was present secondary to the ERM. We conclude that some individuals may have isolated, unilateral, foveal hypoplasia and that an ERM forming over such a fovea may be misinterpreted as having caused macular edema.

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