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Optical Coherence Tomography Angiography Findings in Isolated Foveal Hypoplasia

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Purpose: To describe optical coherence tomography angiography (OCTA) findings in two cases of isolated foveal hypoplasia.

Case summary: (Case 1) An eight-year-old girl presented with amblyopia and exotropia. Optical coherence tomography demonstrated no central concavity in the foveal area with inner retinal layers present. OCTA revealed that lower central retinal vessel density was lower than peripheral retinal vessel density in the deep vascular plexus but not in the superficial plexus. (Case 2) A 14-year-old boy presented with absence of foveal reflex in both eyes. On OCTA, retinal vessel density of the superficial plexus was similar throughout the macula, while lower retinal vessel density was noted in the foveal region than in the parafoveal region of the deep plexus.

Conclusions: OCTA is useful for characterizing foveal hypoplasia. Furthermore, the disparity between deep and superficial retinal vessel density supports the theory of arrested foveal development, in which displacement of deep retinal tissue precedes inner retinal displacement.

Keywords: Foveal development; Foveal hypoplasia; Optical coherence tomography angiography; Retinal vessel density

Introduction

Foveal hypoplasia refers to a condition in which the foveal depression is not formed and all neurosensory retinal layers are observed in the area presumed to represent the fovea [1-4]. Foveal hypoplasia is associated with aniridia, albinism, microphthalmus, achromatopsia, and retinopathy of prematurity; however, it may also occur in isolation [1-3,5,6]. Development of the foveal pit begins at fetal week 25, and is completed by the fourth year after birth [7]. Arrested development may lead to foveal hypoplasia, but the exact mechanism of this remains unclear.

The fovea has a specific region known as the foveal avascular zone (FAZ) that lacks blood vessels. Fluorescein

angiography has revealed that the FAZ is missing in foveal hypoplasia [6]. However, the two distinct layers of the vascular plexus – deep and superficial [8] – of the retina cannot be visualized using fluorescein angiography, but are visible on optical coherence tomography angiography (OCTA). In this report, we describe the retinal vascular plexus using OCTA in two cases of isolated foveal hypoplasia.

Case Report

Case 1

An eight-year-old girl presented with a history of persistent amblyopia in the left eye after strabismus surgery. The pa-

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Figure 1. Fundus examination, optical coherence tomography (OCT) angiography, and OCT B-scan findings in a patient with left foveal hypoplasia (case 1). (A) Fundus appearance of an 8-year-old girl with left isolated foveal hypoplasia. A weak foveal reflex was observed in the left eye. (B) OCT angiography imaging of the superficial retinal plexus of the left eye. White asterisk correspond to the fovea in the OCT B-scan. No foveal avascular zone was observed. (C) OCT angiography imaging of the deep retinal plexus in the right eye. White asterisk also correspond to the fovea on OCT B-scan and yellow arrowheads are projection artifacts of superficial retinal vessels. A partial loss of the deep vascular network in the region presumed to represent the fovea was observed. (D) Horizontal OCT B-scan findings of left idiopathic foveal hypoplasia. The patient's left eye had a shallow foveal pit, and both outer nuclear layer widening and outer segment lengthening were observed. (E, F) Angio B-scan images show the segmentation for the superficial and deep retinal plexus.

tient had undergone left lateral rectus recession and inferior oblique anteriorization due to persistent exotropia and left hypertropia seven months prior. Best corrected visual acuity (BCVA) was 20/20 in the right eye and 20/100 in the left; intraocular pressure (IOP) was 13 mmHg in both eyes. Fundus examination revealed that the foveal reflex was weak in the left eye (Fig. 1A). On OCTA, the FAZ was apparent in both the superficial and deep retinal vascular plexus of the right eye. In contrast, an area of decreased retinal vascular density (RVD) indicating the FAZ was not observed in the superficial plexus, with no difference between the central and peripheral macula in the left eye (Fig. 1B). A partial reduction of RVD was noted in the deep retinal plexus in the region of the fovea (Fig. 1C). On optical coherence tomography (OCT, DRI OCT-1 Atlantis, Topcon Medical, Oakland, NJ, USA), the foveal depression was observed to be normal with the inner retinal layers absent in the right eye. In the left eye, the foveal concavity was blunted with preservation of the ganglion cell

layer (GCL), inner plexiform layer (IPL), and inner nuclear layer (INL) (Fig. 1D). The outer nuclear and plexiform layers (ONL and OPL, respectively) were thickened in the presumed foveal area, and no obvious abnormalities of the photoreceptor layer were observed in either eye (Fig. 1E, F).

Case 2

A 14-year-old boy was referred with an incidental abnormal finding on fundus examination. No associated medical history was identified. BCVA of the right (Fig. 2A-F) and left (Fig. 2G-L) eyes was 20/20 and 20/25, respectively, and IOP was 14 and 16 mmHg, respectively. On fundus examination, no foveal reflex was observed in either eye (Fig. 2A, G). On OCTA, RVD was not found to be reduced, and involvement of large-caliber vessels in the central area was observed in the superficial plexus (Fig. 2B, E, H, K). A partial reduction in RVD was observed in the deep retinal vascular plexus (Fig.

Figure 2. Fundus examination, optical coherence tomography (OCT) angiography, and OCT B-scan findings in a patient with bilateral foveal hypoplasia (case 2). White asterisk correspond to the fovea in the OCT B-scan. (A) Fundus appearance of a 14-year-old boy with isolated foveal hypoplasia. No foveal reflex was observed in the right eye. (B) OCT angiography imaging of the superficial retinal plexus revealed no reduction in retinal vascular density in the central macula. (C) OCT angiography of the deep retinal plexus demonstrates a partial loss of deep retinal vessels in the central macular area. (D) The patient had no foveal pits, but there were no obvious abnormalities in the photoreceptor layer. (E, F) Angio B-scan images depict the segmentation for the superficial and deep plexus in panels B and C, respectively. (G-L) The left eye exhibited similar findings on fundus examination, OCT angiography and OCT B-scan.

2C, F, I, L). OCT B-scan demonstrated loss of the foveal depression in both eyes, and discontinuities were not observed in the inner retinal layers of the GCL, IPL, and INL. The ONL and OPL were thickened in the presumed foveal region, whereas the outer segment (OS) of the photoreceptor layer was not (Fig. 2D, J).

Discussion

Foveal hypoplasia is characterized by decreased foveal reflex on fundus examination [6]. Our two cases exhibited typical characteristics of foveal hypoplasia, including preservation of the inner retinal layers on OCT. Investigations using OCTA revealed that the FAZ was not present in the superficial plexus but was partially developed in the deep plexus. These observations are consistent with previous reports [9,10] and indicate arrested development of the fovea.

Thomas et al. [11] proposed a structural grading system of foveal hypoplasia based on anatomical differences seen on OCT. Four grades of foveal hypoplasia were identified and correlated with visual acuity. The authors suggested that the fovea developed in the following order: centripetal migration of the cone and ONL widening, OS lengthening, formation of the foveal pit, and extrusion of the plexiform layers. Arrest at an early stage results in a more severe grade of foveal hypoplasia and worse visual acuity. However, Marmor et al. [6] reported four cases of foveal hypoplasia with visual acuity ranging from 20/20 to 20/50, and claimed that the foveal pit is not required for good visual acuity. They suggested the new term for this condition, fovea plana, instead of foveal hypoplasia, which implies poor function.

In case 1 of this report, the patient's left eye had a shallow foveal pit, ONL widening, and OS lengthening, corresponding to the grade I classification proposed by Thomas et al. [11] Despite strabismus surgery and occlusion therapy, visual

acuity remained low. On the other hand, both eyes in case 2 were classified as grade 3, showing only ONL widening with no foveal pit or OS lengthening, but visual acuity was 20/20 and 20/25 in the right and left eyes, respectively. Therefore, in our patients with foveal hypoplasia, visual acuity had no clear correlation with anatomical grading. Our results are more in line with those of Marmor et al. [6] than with those of Thomas et al. [11].

The grading system of Thomas et al. [11], however, explains the development of the fovea in detail. On OCTA, our patients had some loss of RVD in the deep plexus but not in the superficial retinal vascular plexus. The current OCTA findings are consistent with the developmental process occurring in the outer layer before the inner layer, and suggest that the formation of the FAZ is not complete before extrusion of the plexiform layers.

In summary, a novel imaging technique, OCTA, can be used to visualize the deep and superficial retinal plexus separately, and will be helpful in the diagnosis and classification of foveal hypoplasia when combined with conventional OCT B-scan. The disparity between the deep and superficial RVD suggests that displacement of the deep retinal plexus precedes displacement of the superficial retinal plexus. Further investigation of the complex relationship between anatomical findings and visual function is needed.

Conflicts of interest

The authors have no conflicts to disclose.

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