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Unilateral isolated foveal hypoplasia

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Unilateral isolated foveal hypoplasia

A 9-year-old male was referred to the Pediatric Ophthalmology service for a suspected epiretinal membrane at the left macula. The patient was asymptomatic. His ocular, systemic medical, birth, and developmental history were all noncontributory, and he was not on any prescription medication.

On examination, his best-corrected visual acuity was 6/6 OD and 6/9 OS. Posterior pole examination of the left eye revealed a diminished foveal reflex and smaller foveal region with a less prominent mound. Furthermore, a retinal artery was seen coursing through the papillomacular bundle. The optic nerves and peripheral retina in both eyes were normal, as was the posterior pole in the right eye (Fig. 1). His ocular motility, alignment, and anterior segment examination results were all within normal limits. There was no aniridia or nystagmus, and there were no iris transillumination defects. Cycloplegic refraction revealed a refractive error of +1.00 in either eye (Fig. 2).

An optical coherence tomography (OCT) confirmed the clinical suspicion of a grade 1 foveal hypoplasia isolated to the left eye. Owing to the absence of other ocular findings and other systemic pathology, the prognosis was thought to be favourable, and we decided that no further work-up was indicated at that time.

Discussion

Foveal hypoplasia (FH) refers to underdevelopment of the fovea, characterized by the continuity of inner retinal layers in the presumed foveal area; the foveal avascular zone may be limited or absent, and retinal vessels may be seen coursing through the foveal region.[1,2] It is commonly associated with ocular conditions such as aniridia,

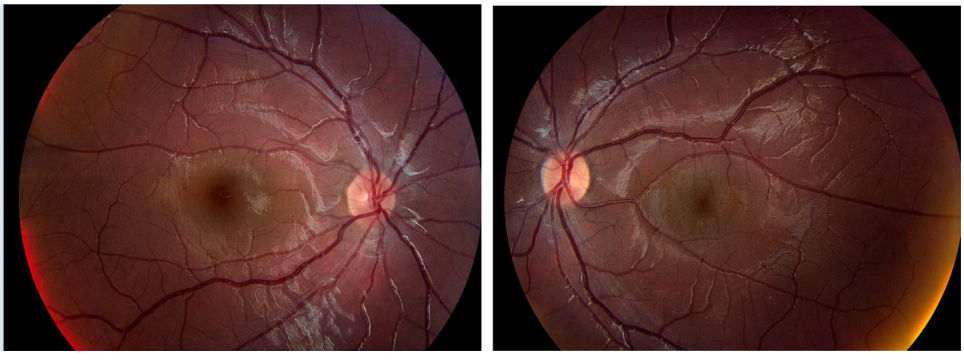

Fig. 1—Colour fundus photographs displaying normal anatomy in the right eye and abnormal distribution of retinal vasculature and smaller diameter and elevation of foveal mound in the left eye.

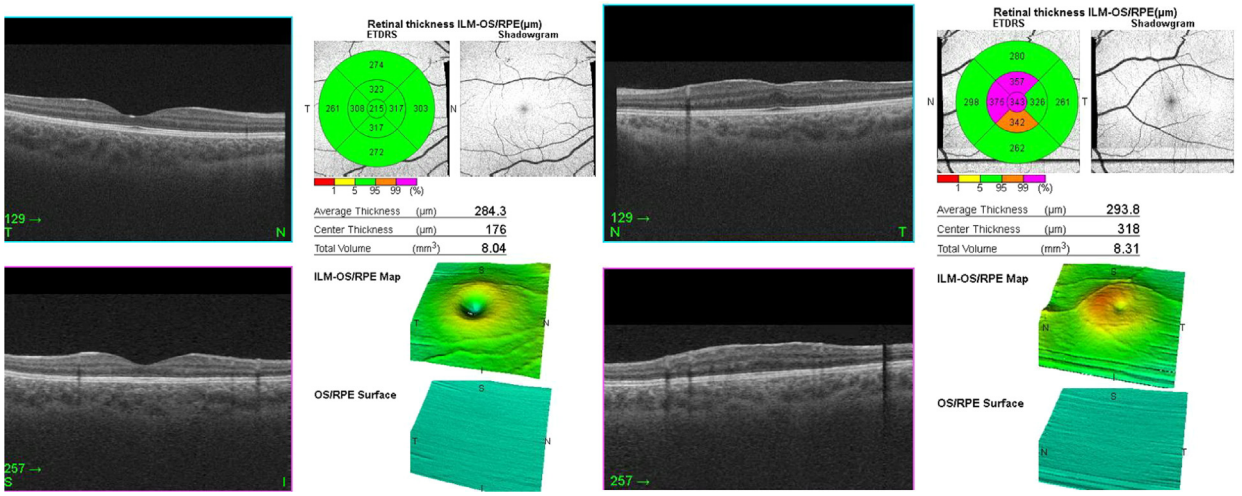

Fig. 2—Optical coherence tomography of both eyes, showing normal anatomy in the right eye and persistence of inner retinal layers and loss of foveal depression in the left eye.

albinism, achromatopsia, or a PAX6 mutation.[3] Less commonly, isolated foveal hypoplasia (IFH) may occur. All but one[4] case of IFH that have been described are bilateral and are associated with varying degree of vision loss, nystagmus, and typical ocular findings.

Normal foveal development occurs gradually—beginning at 25 weeks of gestational age, complete foveal pit formation can continue until 15 months of age, and refinement of cone positioning and size can continue up to 45 months of age. The foveal region remains avascular throughout development, and failure of this avascularity to persist is thought to be a precursor to FH.[5] Three steps[6] are essential to foveal development; anatomical progression is visualized quite well with high-definition OCT. The steps are as follows: (*i*) centrifugal displacement of inner retinal cells allows for the foveal depression to begin and, by the time of completion, is seen on OCT as complete extrusion of inner retinal layers; (*ii*) centripetal migration of cones, which is represented on OCT as outer nuclear layer (ONL) widening; (*iii*) specialization of the foveolar cones, wherein the cone outer segments undergo a decrease in diameter and an increase in length, allowing for a denser packing of cone cells at the fovea, which is represented on OCT as outer segment (OS) lengthening.

The diagnostic criteria for FH is limited to the continuity of inner retinal layers. As such, there is significant phenotypic variability in foveal anatomy and visual acuity. A structural grading scheme looking at the anatomy and visual acuity has been published[6] and described in detail. Briefly, the 4 grades of FH are based on deviation from the aforementioned norm and are as follows: (*i*) persistence of inner retinal layers, (*ii*) absence of a foveal pit, (*iii*) absence of outer retinal lengthening, and (*iv*) absence of ONL widening. Furthermore, if there is disruption of the IS/OS junction, which is uncommon in IFH but seen more frequently in FH associated with achromatopsia, it is deemed "atypical" foveal hypoplasia.

As the structure relates to function, the relationship between foveal depression and visual acuity is controversial. Thomas et al.[6] found that, regardless of the diagnosis, there was a significant relationship between best-corrected visual acuity and the grade of hypoplasia. Grade 1 hypoplasia was associated with the best visual acuity, with incremental decline in visual acuity with grades 2, 3, and 4 hypoplasia, respectively. Having said this, others hypothesize that visual prognosis has more to do with macular pigmentation and foveal cone specialization than with the foveal depression.[3,7] Although Thomas et al.'s report seems promising in terms of offering a predictor of visual acuity, the consensus in the literature to date remains inconclusive regarding the form–function relationship in FH.

Delving further into structural changes in FH, it is logical to look next into the differences in the dimensions as seen on OCT. Although this particular area has not been thoroughly investigated, Holmström et al.[8] have looked at OCT images of patients with FH secondary to aniridia and albinism and compared those dimensions to patients with normal maculae. Although they did not look at IFH, the relevant findings in this work were that the thinnest point of the fovea was thicker in the patients with FH, as was the thickness of the central 1 mm of the macula. These were both reproduced in our case—the thinnest point in the eye with FH was 318 µm and the central 1 mm measured 343 µm. These were both thicker than that in the patient's normal eye, in whom the values measured 176 and 215 µm, respectively. Furthermore, these values in the normal eye correspond to the normative OCT macular data in the eye of healthy children,[9] whereas these same values in the eye with FH fall outside of this range.

Although IFH is an uncommon diagnosis, the subtle examination findings and the significant phenotypic and functional variability make it quite plausible that it is underdiagnosed.[10]

Conclusion

We present a very rare case of unilateral isolated foveal hypoplasia diagnosed in a 9-year-old male. Our case shows persistence of inner retinal layers with a shallow foveal depression, which, based on the above classification, constitutes grade 1 isolated unilateral foveal hypoplasia.

Disclosure: The authors have no proprietary or commercial interest in any materials discussed in this article.

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Mantle cell lymphoma: conjunctival mass in a female patient

Mantle cell lymphoma (MCL) is a rare neoplasm in the ocular adnexa, accounting for 3%–5% of conjunctival B-cell non-Hodgkin lymphomas (NHL).[1,2] Conjunctival MCL typically presents in male patients in the eighth decade and is often advanced at the time of presentation.[3] Treatment typically consists of chemotherapy or radiation, although the prognosis is poor.[4] The authors present a rare case of conjunctival MCL in a female patient. Collection and evaluation of protected patient health information complied with the Health Insurance Portability and Accountability Act.

Case Report

A 61-year-old female presented with a painless, salmon-colored conjunctival lesion approximately 8 × 5 mm in size in the medial canthal region of the right eye (Fig. 1). The mass had been present for 2 weeks, and at her follow-up visit 1 month later it appeared to have grown. She had no visual symptoms or complaints in the right eye, although she had been experiencing flashes and floaters in the left eye, which was found to be a posterior vitreous detachment. Ophthalmic examination was otherwise within normal limits. No palpable or visible posterior extension could be appreciated. Patient denied B-symptoms such as fever, night sweats, or lymphadenopathy. She had no history of malignancy.

There was high clinical suspicion of lymphoma, and the patient subsequently underwent biopsy of the right conjunctival lesion. Histopathology showed soft tissue infiltration of fairly uniform atypical lymphocytes with slightly irregular nuclear membranes and focally vesicular chromatin without prominent nucleoli. Occasional mitoses were

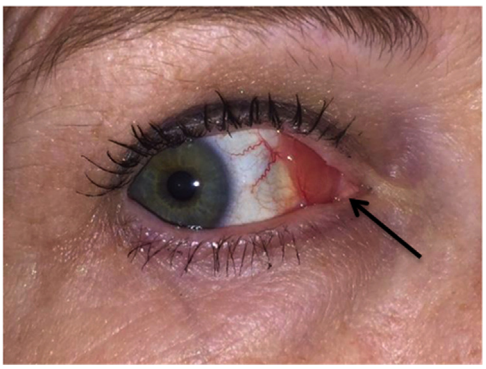

Fig. 1—Right eye in abduction showing a salmon-colored conjunctival lesion near the medial canthus, approximately 8 × 5 mm in size.