

OCT Findings in A Case With Iris Coloboma and Foveal Hypoplasia

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ABSTRACT

In foveal hypoplasia there is a continuity of all neurosensory retinal layers in the fovea and loss of foveal pitting. It can be observed in patients with albinism, microphthalmia, aniridi, acromatopsia and premature retinopathy, and rarely in isolated cases. In our case, who presented to our clinic with complaints of deviation in the eyes and decreased distance vision, iris coloboma was present in both eyes on anterior segment examination. On fundus examination, foveal reflex was absent and retinal vessels were observed as slightly tortuous in both eyes. There was no foveal pitting on OCT sections in both eyes. With these findings our case was diagnosed with foveal hypoplasia. The aim of this article is to present the case of iris coloboma and foveal hypoplasia, in whom OCT have an important place in the diagnosis, follow-up and classification.

Keywords: Foveal hypoplasia, iris coloboma, OCT.

INTRODUCTION

Foveal hypoplasia is defined as lack foveal pitting together with continuity of all neurosensory retinal layers in fovea¹. It is known that foveal hypoplasia can be seen in patients with albinism, microphthalmia, aniridia, achromatopsia and retinopathy of prematurity while it can be seen as isolated entity in rare instances². Currently, several macular and retinal diseases can be readily detected by optical coherence tomography (OCT). The detection and grading of foveal hypoplasia can be readily achieved using OCT by foveal development defect³.

Here, it was aimed to describe a patient with iris coloboma and foveal hypoplasia.

CASE REPORT

A 6-years old girl presented to our clinic with esophoria and impaired distance acuity. There was no history of systemic disease or medication in the patient. There was history of preterm birth (2 weeks before) and consanguinity between parents. In ophthalmological examination, best-corrected visual acuity was 0.7 (+2.50 +1.00x 25) in right eye and 0.4 (+2.50 +1.00x155) in the left eye. In the strabismus examination, there was esotropia at primary position with

25 PD at near and 18 PD at distance in alternate prism cover test. Ocular movements were normal in all directions without nystagmus. In anterior segment examination, cornea and lens was normal in both eyes and there was a defect compatible with coloboma in inferior quadrant of iris (Figure 1A-B). On fundus examination, optic disc was normal in the right eye while foveal reflex was absent and retinal vessels were mildly tortuous (Figure 2A-B). In the left eye, optic disc was normal while there was myelinated nerve fiber at superior and nasal regions of optic disc and foveal reflex was absent with tortuosity in retinal vessels (Figure 3A-B). Foveal scans were obtained by time-domain optical coherence tomography (TD-OCT Stratus OKT, Carl Zeiss Meditec, USA). It was seen that foveal pitting was absent in both eyes on OCT (Figure 4A-B and 5A-B). In detailed assessment, it was seen that, in addition to absence of foveal pitting, there was no disruption at inner and outer plexiform layer; that there was no elongation of photoreceptor outer segment; and that outer nuclear layer continued to enlarge. Based on these findings, the patient was assessed as bilateral, stage 3 foveal hypoplasia. The parents were informed about condition and the patient was scheduled to follow-up with eye occlusion treatment for amblyopia.

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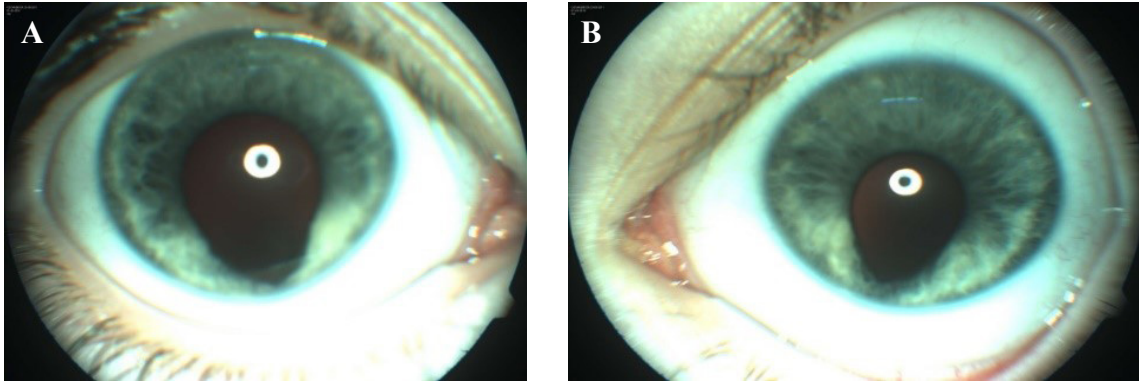


Figure 1A-B: Anterior segment view of right and left eyes.

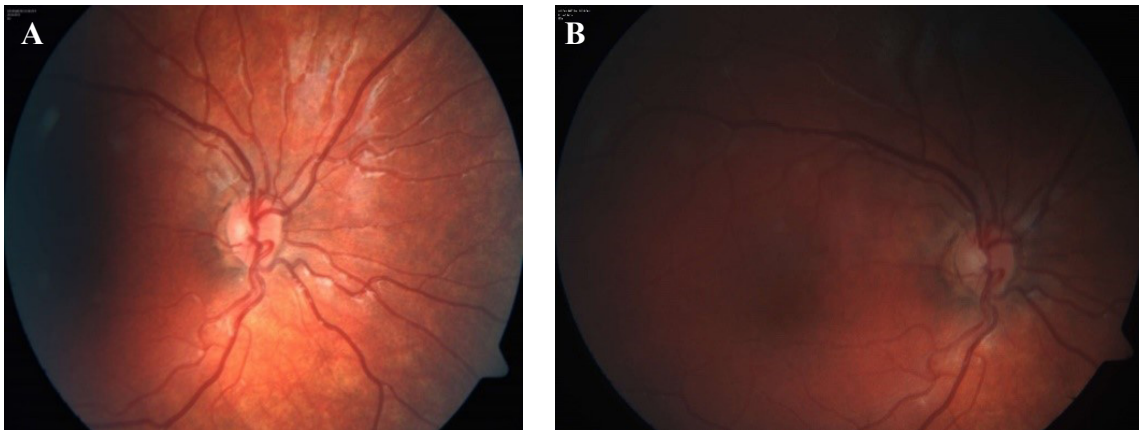


Figure 2A-B: Fundus image of right eye.

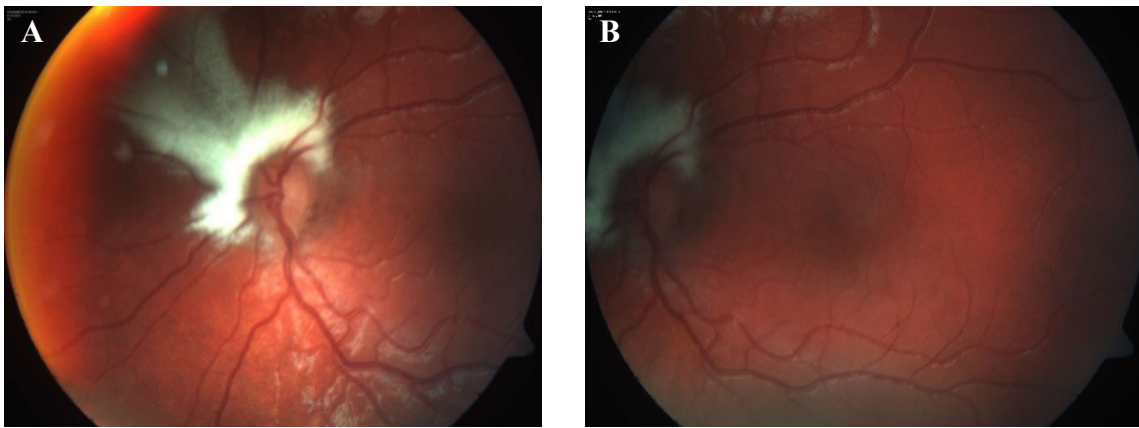


Figure 3A-B: Fundus image of left eye.

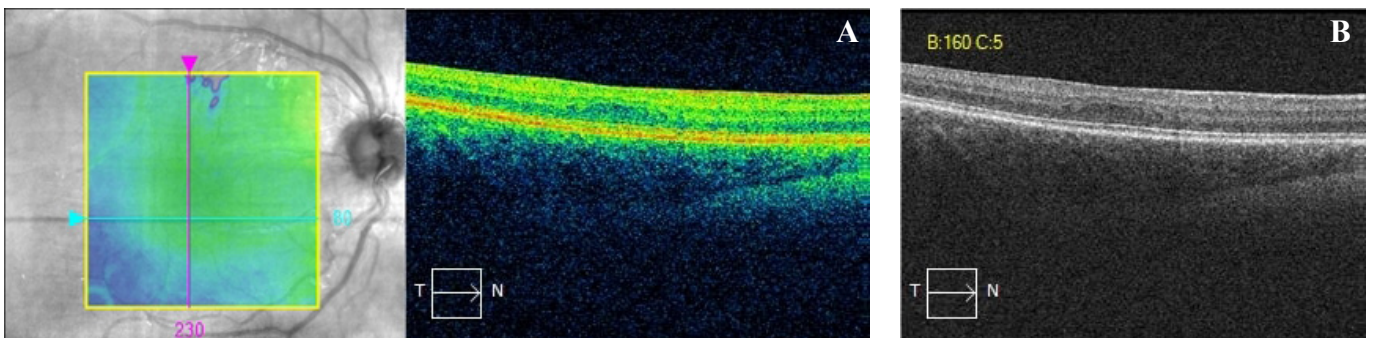


Figure 4A-B: OCT image of right eye: no foveal pitting; inner retinal layers continue at fovea; no elongation in cone outer segment; enlargement in outer nuclear layer.

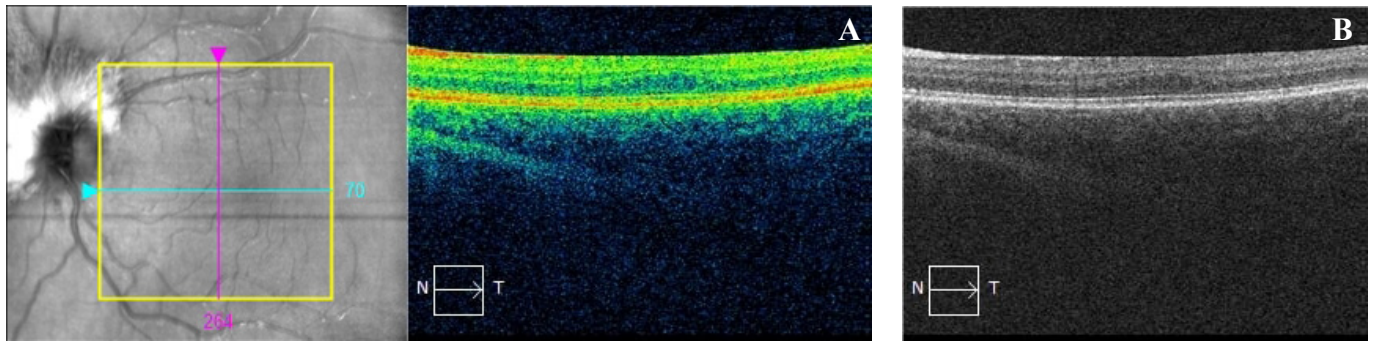


Figure 5A-B: OCT image of left eye: no foveal pitting; inner retinal layers continue at fovea; no elongation in cone outer segment; enlargement in outer nuclear layer.

DISCUSSION

In 1984, Hendrickson and Yuodelis proposed that isolated foveal hypoplasia is halt in foveal development stages during fetal period⁴.

The association of foveal hypoplasia with albinism, aniridia, retinopathy of prematurity and incontinentia pigmenti is well-known. However, autosomal dominant isolated foveal hypoplasia with no comorbid ocular pathology has also been described in the literature. By introduction of OCT, it was shown that foveal hypoplasia can accompany achromatopsia, optic nerve hypoplasia, familial exudative vitreoretinopathy and Stikler syndrome⁵.

In 2011, Thomas et al. assessed foveal hypoplasia by classifying in 4 grades according to OCT findings. Authors classified foveal hypoplasia into 4 grades based on finding such as presence of inner retinal layers in fovea, absence of foveal pitting, elongation of photoreceptor outer segment and enlargement of outer nuclear layer on OCT. Accordingly, the inner nuclear and inner-outer plexiform layers are present posterior to fovea with shallow foveal pit in grade 1. While foveal pit is lacking in grade 2. The foveal pit and outer segment elongation are lacking in grade 3. In grade 4, the thickening at outer nuclear layer is lacking in addition to finding in grade 3⁶. In our case, it was found that the foveal pit was lacking in both eyes and inner and outer plexiform layers continued in fovea but there was no elongation in photoreceptor outer segment. However, it was seen that outer nuclear layer enlargement continued. Based on these finding, the patient was considered to have bilateral, grade 3 foveal hypoplasia. The absence of foveal reflex was evaluated using OCT and decreased vision was attributed to strabismus and fovea hypoplasia. Although there is no study on amblyopia in foveal hypoplasia, it was shown that amblyopia treatment can be successful in optic nerve hypoplasia⁷. Our case was scheduled for follow-up with treatment of amblyopia.

Schroeder HW et al. defined PAX6 mutation in an autosomal dominant disorder characterized by association

of esotropia, cataract and foveal hypoplasia. Authors suggested that foveal hypoplasia was seen in all cases but iris involvement can be variable. However, foveal hypoplasia seen in albinism can be variable while VEP asymmetry is an universal finding. Thus, authors suggested that it is possible to determine whether foveal hypoplasia is due to aniridia or albinism by VEP⁸. In our case, there was no cataract, finding of albinism and nystagmus. In addition, the patient could not classified into a known disease group since no genetic testing was performed. We evaluated the macula in details using OCT.

CONCLUSION

The OCT is a practical and reliable method for diagnosis and classification of foveal hypoplasia. We think that foveal hypoplasia will be more commonly detected and classified by widespread use of OCT with above-mentioned clinical characteristics.

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