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ISOLATED FOVEAL HYPOPLASIA: A CASE REPORT

Biljana Ivanovska Adjievska[1,2], *Violeta Buckoska[1]*

[1]"Denica" Specialised Eye Hospital, Skopje, North Macedonia;
[2]Faculty of Medical Sciences, Goce Delcev University, Stip, North Macedonia
e-mail: ivanovska.biljana@gmail.com, violetabuckoska@gmail.com

Abstract: Foveal hypoplasia is a retinal disorder in which the foveal pit of the macula lutea is incompletely developed and is characterized by nystagmus and low visual acuity. It can manifest itself in isolation, without a clear etiology, or associated with other conditions such as albinism, aniridia, Stickler Syndrome, optic nerve hypoplasia, microphtalmus, etc. Modern retinal imaging technologies are key to diagnosis of this rare retinal disorder. We present a case of a 19-year-old woman with poor distance and near vision and nystagmus since childhood, without other associated diseases. Visual acuity was 0.5 Snellen decimal units in both eyes, with present latent nystagmus. The anterior segment was normal, while an absent foveal reflex was found on fundus examination. Optical coherence tomography (OCT) imaging confirmed the absence of foveal depression in the macular area. OCT-angiography (OCTA) was performed, which confirmed the absence of a foveal avascular zone in the macula. According to the proposed grading system parameters for foveal hypoplasia, this case corresponds with the most severe (grade 4) degree of foveal hypoplasia. Foveal hypoplasia can be associated with numerous etiological factors, and visual acuity can vary depending on the development of foveal photoreceptors and structural gradation. For this reason, especially in children with reduced visual acuity of unknown etiology and the existence of nystagmus, it is recommended to perform additional examinations and use multimodal imaging techniques (OCT and OCTA) in order to make a timely and accurate diagnosis. Management of this disorder includes treatment of the associated ocular and systemic conditions, refractive correction, treatment of amblyopia and use of low vision aids.

Keywords: Foveal hypoplasia, nystagmus, optical coherence tomography
Field: Medical sciences and Health, Ophthalmology

INTRODUCTION

Foveal hypoplasia is a retinal disorder in which the foveal pit of the macula develops only partially or not at all and is characterized by nystagmus and low visual acuity. It can present in isolation, without a clear etiology, or be associated with other conditions such as albinism, optic nerve hypoplasia, aniridia, familial exudative vitreoretinopathy (FEVR), retinopathy of prematurity, Stickler syndrome and microphthalmia. A study on healthy children by Noval et al., 2014 found that up to 3% of children had an anatomically underdeveloped foveal pit bilaterally on OCT. In patients with foveal hypoplasia the visual acuity varies from 0.1 to 0.6 Snellen decimal units and they lack foveal depression and pigmentation. Several studies (Kuht et al, 2022; Ehrenberg et al., 2021) have established an association of foveal hypoplasia with mutations in genes associated with albinism (PAX6, GPR143, OCA2, etc.)

Optical coherence tomography (OCT) of a normal fovea shows a central depression with loss of the inner retinal layer and elongation of the photoreceptor outer segments. In addition, OCT angiography (OCTA) records that the fovea has a central avascular zone, which is absent in cases of foveal hypoplasia. In 2020 Rufai SR et al. developed a staging system for patients with foveal hypoplasia, was based on the morphological characteristics detected by OCT.

In our study, we present a case of isolated foveal hypoplasia, confirmed by OCT and OCT angiography.

CASE REPORT

A 19-year-old woman was examined in our hospital, who complained of poor distance and near vision and presence of nystagmus since childhood. Anamnestically, the patient did not report any other more serious systemic diseases, nor family history of poor vision or nystagmus.

Ophthalmological examination revealed a visual acuity of 0.5 Snellen decimal units in both eyes, which did not improve with the existing refraction (in cycloplegia: OD: +0.75 Dsph -0.75 Dcyl. ax. 175, OS: +0, 75 Dsph -1.00 Dcyl. Ax. 5). IOP was normal (TOD: 13 mmHg, TOS: 10 mmHg). The patient had latent

*Corresponding author: ivanovska.biljana@gmail.com

nystagmus, but no manifest or latent strabismus was observed.

Biomicroscopic examination of the anterior segment was normal, with no indications of ocular albinism, such as transillumination, and iris pigmentation was normal. Fundus examination revealed a normal finding of the optic nerve head and blood vessels, but absent foveal reflex and pigmentation in the macula.

An OCT image was performed (Figure 1), which revealed the absence of foveal depression in both eyes, with preserved continuity of the inner retinal layers. The outer segments of the photoreceptors were not extended, and minimal expansion of the outer nuclear layer was recorded.

On the performed OCT angiography (Figure 2), the absence of a foveal avascular zone in the superficial capillary plexus was observed, as well as marked reduction in the deep capillary plexus.

The patient was informed of the diagnosis and suggested to undergo genetic analyzes for the presence of possible gene mutations associated with this disorder.

Figure 1. Optical coherence tomography of the right and left eye shows absence of foveal depression.

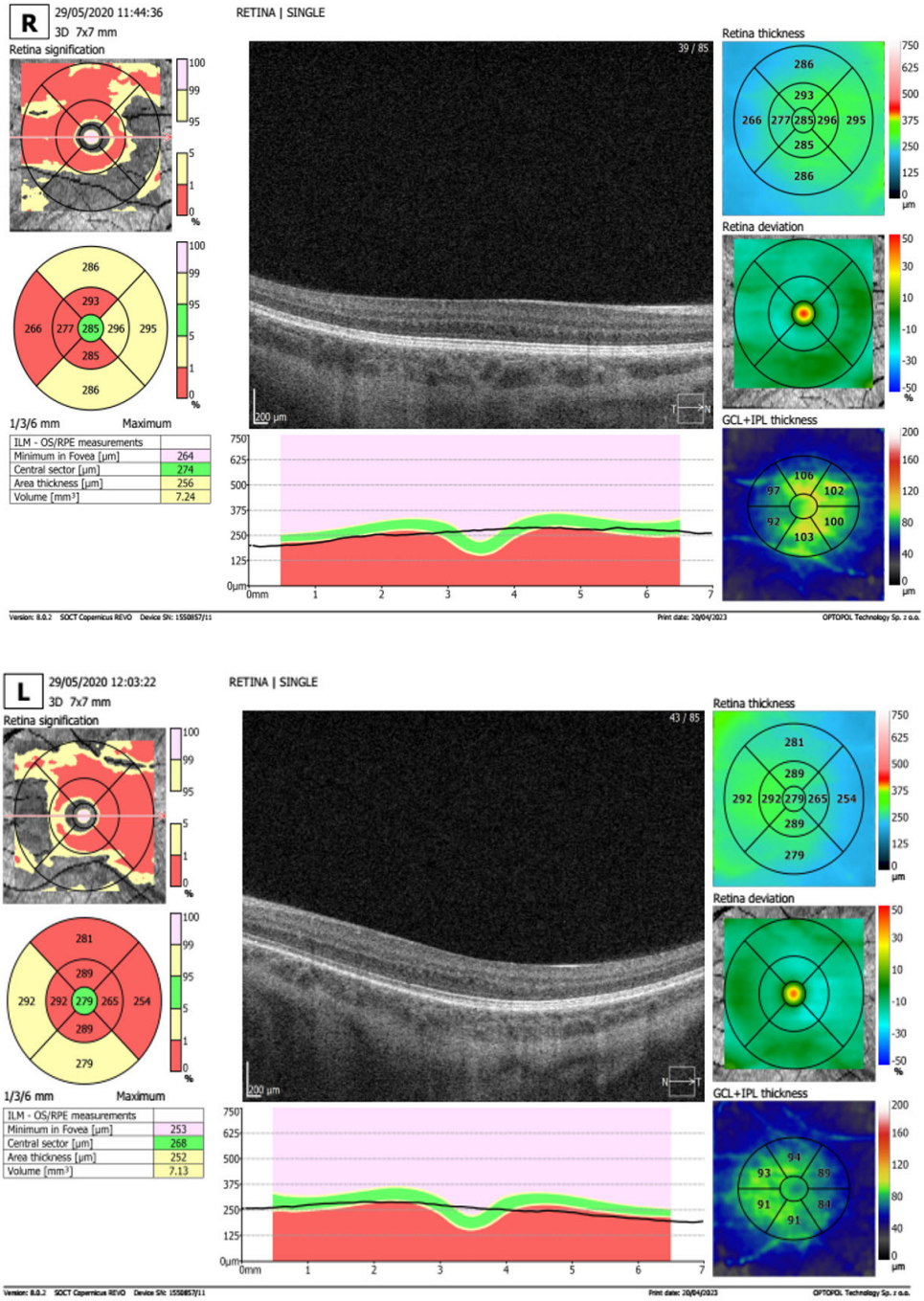
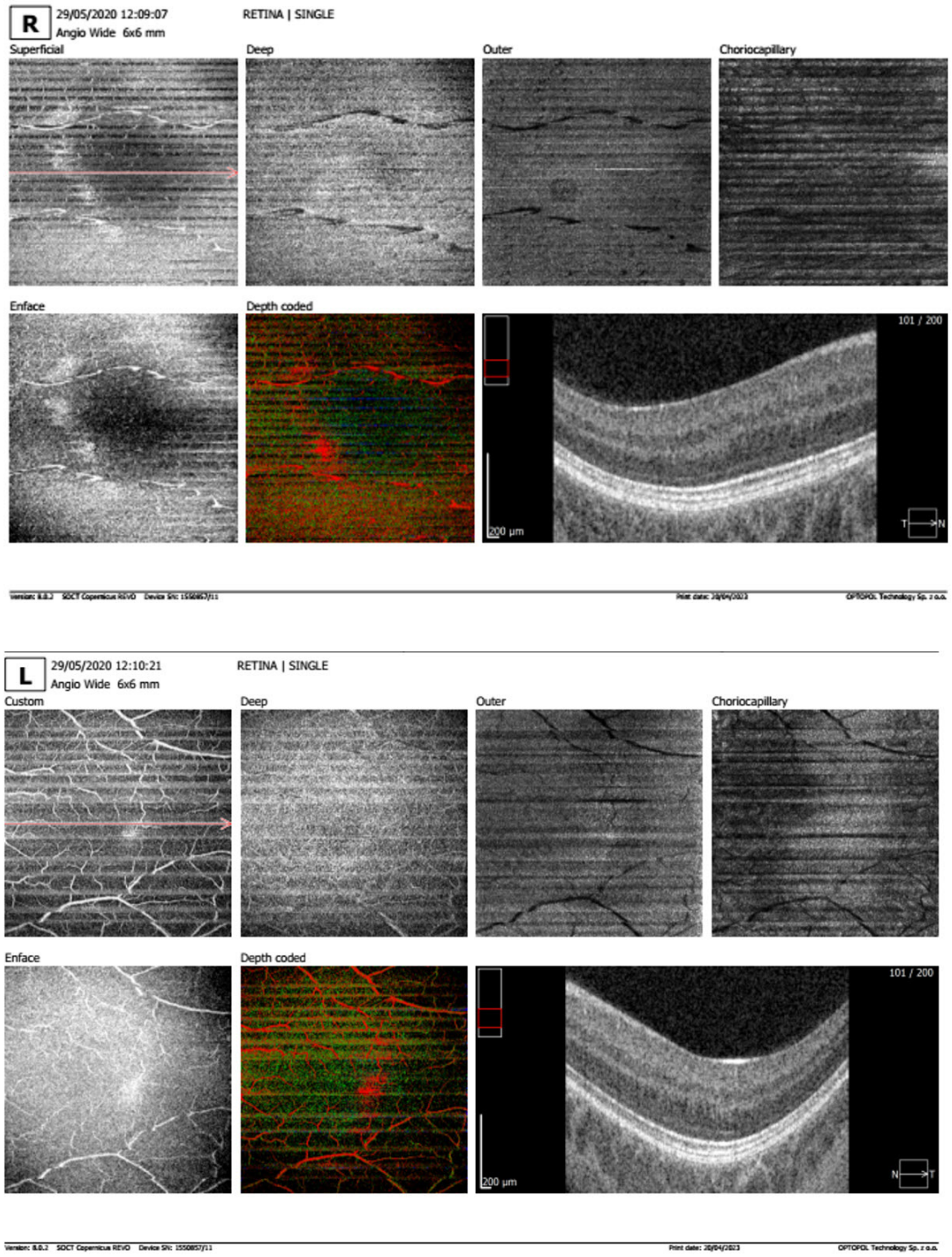


Figure 2. OCT angiography of the right and left eye shows absence of foveal avascular zone.



DISCUSSION

In our study, the patient had isolated foveal hypoplasia associated with underdeveloped fovea and nystagmus, but without associated etiology (albinism, aniridia, optic nerve hypoplasia, FEVR, Stickler syndrome, microphthalmia, nanophthalmus, achromatopsia, etc.).

Visual acuity is generally reduced in patients with foveal hypoplasia. Rufai et al., 2020 proposed a system for categorizing foveal hypoplasia in order to predict visual acuity based on OCT scans. The authors proposed 3 key points for classification: the degree of foveal depression, elongation of the outer segment of the photoreceptors, and expansion of the outer

nuclear layer. In our case, the patient has no foveal pit, no elongation of the outer segment of the photoreceptors and only a slight expansion of the outer nuclear layer, which corresponds to the worst prognosis, i.e. 4th degree of foveal hypoplasia. According to the mentioned classification, the visual acuity would be expected to be 0.1-0.2, however, it was 0.5 Snellen decimal units in the patient. Therefore, it is possible that the visual acuity and the structural characteristics of the fovea are not correlated, which requires further investigations.

OCT angiography is a simple, fast and non-invasive method for imaging the retinal blood vessels in the macular area. In foveal hypoplasia, it can reveal the lack of a foveal avascular zone in the superficial and deep capillary layers (Pakzad-Vaezi et al., 2017; Sánchez-Vicente et al, 2018). In our case, the patient had the absence of a foveal avascular zone in both eyes.

CONCLUSION

Foveal hypoplasia can be associated with numerous etiological factors, and visual acuity can vary depending on the development of foveal photoreceptors and structural gradation. Modern imaging technologies (OCT and OCT angiography) are crucial in diagnosing this disorder. Therefore, especially in children with reduced visual acuity of unknown etiology and the existence of nystagmus, it is recommended to perform additional multimodal imaging examinations in order to make timely and accurate diagnosis.

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