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Rare presentation of foveal hypoplasia: A case report

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Abstract:

Twenty-five years lady presented to retina outpatient department for routine check-up. She gave history decrease in vision both eyes since childhood. Her vision on examination was 2/60 in both eyes with no improvement with glasses. On examination there was nystagmus, anterior chamber depth, iris colour, lens and vitreous were normal. On fundus examination, optic disc, retinal vessels were normal. There was no foveal reflex and lattice degeneration was noticed superotemporally. Optical coherence tomography confirmed features of atypical foveal hypoplasia both eyes

Keywords: Foveal hypoplasia, Lattice degeneration, Preterm

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Introduction

Normal development of the fovea begins in utero at 12th fetal week, and has 3 stages, 1st centrifugal displacement of cells of the inner retina towards the periphery resulting in fovea depression seen as complete extrusion of the inner nuclear and plexiform layers posterior to the foveola. In 2nd stage the centripetal migration of cone photoreceptors towards the location of the incipient fovea results in outer nuclear layer (ONL) widening. In 3rd stage the cone outer segment decreases in diameter and increase in length resulting in cone specialization resulting in increase in foveolar cone packing density. The change in cone diameter continues up to 45 months postpartum. The cone specialization is represented on optical coherence tomography by the Outer segment (OS) lengthening.¹

Foveal hypoplasia is an ocular abnormality in which the foveal pit either fails to develop, or does not completely develop, and is associated with poor visual acuity and nystagmus.¹ It may present in isolation or with albinism, coloboma, optic nerve hypoplasia, retinopathy of prematurity or aniridia.² Prematurity is a significant risk factor for abnormal development of retinal vasculature and deficiency of macular pigmentation.³ Foveal hypoplasia may be related gene mutations of PAX6, OCA2 and GPR143 that are associated with albinism.⁴ Histologic analysis and optical coherence tomography (OCT) studies have revealed that the structure of the macula resembles peripheral retinal tissue with the persistence of ganglion and nuclear layers.⁵

This is rare case report of foveal hypoplasia in a young lady.

Case report

A 25-year lady presented to retina clinic for regular checkup. She gave history of non-progressive low visual acuity since childhood. According to her mother she was delivered premature but was not admitted to neonatal care unit. There was no other significant medical

or surgical history. On examination her eyes, skin and hair colour was brown. There was no mid facial hypoplasia, no hearing loss, joint hypermobility.

On ophthalmic examination, her best corrected visual acuity was 2/60 with no improvement with glasses. There was horizontal nystagmus both eyes. Examination of the anterior segment did not show abnormality. On fundus examination, optic disc and vessels were normal, no foveal reflex and lattice degeneration noticed superotemporally in both eyes.

Spectral domain optical coherence tomography images showed features of loss of foveal pit, loss of extrusion of plexiform layers with continuity of the inner retinal layers and inner segment/outer segment (IS/OS) disruption which were features of atypical foveal hypoplasia in both eyes., Figure 1 and Figure 2.

Discussion

In the current report, we present a patient with atypical foveal hypoplasia associated with nystagmus and history of premature birth. Most of the patients with foveal hypoplasia have decrease in vision. A grading system for foveal hypoplasia to predict the prognosis of visual acuity based on optical coherence tomography scans is widely used in clinical practice.⁵

In the current case, the patient had no foveal depression or lengthening of the outer segment of the photoreceptors; both of which correspond to the worst prognosis. Based on these findings, the patient was diagnosed to have atypical foveal hypoplasia both eyes.

A normal axial length, provided by biometry excluded microphthalmos. Nystagmus was present but iris transillumination was absent which eliminates the probability of ocular albinism. Studies have shown foveal hypoplasia related to retinopathy of prematurity, but she doesn't have any ocular features of retinopathy of prematurity except the lattice degeneration in the periphery.

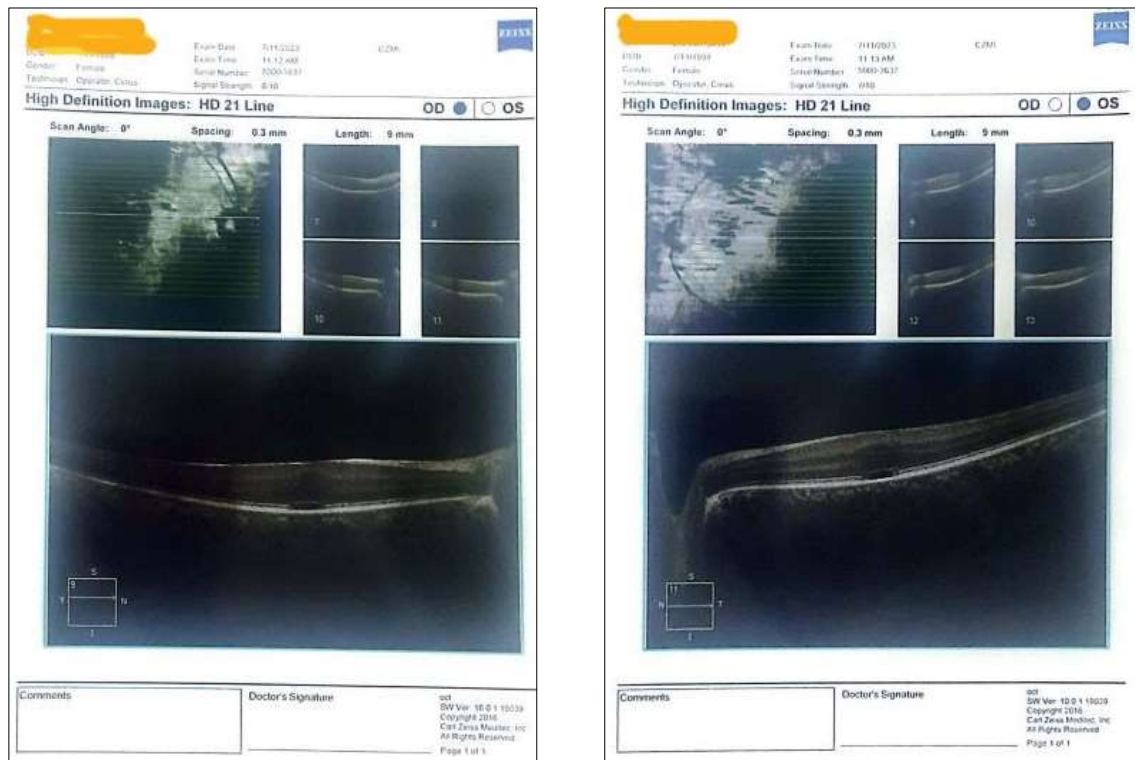


Figure 1 Right eye optical coherence tomography scan of macula showing loss of foveal pit incursion of plexiform layer and disruption of inner segment/outer segment junction; **Figure 2.** Left eye

Conclusion

This is a rare case of atypical foveal hypoplasia both eyes in a lady of 25 years with a history of premature deliver, and decreased vision of 2/60, no foveal reflex or ocular features of retinopathy of prematurity. She had lattice degeneration superotemporally. The optical coherence tomography confirmed and was helpful in diagnosis.

Author contribution

Concept design, Literature search, Data collection, Data analysis, Draft manuscript, Final manuscript and accountability: AM

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Conflict of interest

None

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Consent

All the appropriate patient consent has been taken in the form of verbal and written. In the consent form, the patient gave consent for her images and other clinical information to be reported in the journal. The patient was counselled that her name and initials would not be published and due efforts would be made to conceal the identity, but anonymity cannot be guaranteed.

Supplementary material

The data and supplementary material that support the findings of this study are available from the corresponding author upon reasonable request.

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