Isolated primary foveal hypoplasia

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Introduction
Foveal hypoplasia is a morphological abnormality which represents different stages of arrested development of fovea. It encompasses complete absence (foveal aplasia) or partial absence (foveal dysgenesis) of foveal structure. It is characterized by the absence of foveal pigmentation and/or the foveal avascular zone. It may occur in isolation or in association with other conditions such as aniridia, albinism, achromatopsia, microphthalmos, retinopathy of prematurity (ROP), myopia and incontinentia pigmeni (IP).1–4 The clinical diagnosis might often be missed due to the subtle nature of the fundus findings. This is a case report of an isolated foveal hypoplasia in which optical coherence tomography (OCT) was used to confirm the clinical suspicion of foveal hypoplasia in a 4-year-old boy.

Case Summary
Mother of a 4-year-old boy was referred to the vitreous-retina service of our tertiary eye care hospital with the presenting complaints of child having shaking of the eyeballs since birth with abnormal head posture and with no history of any day or night blindness, photophobia, or premature birth. Parents have a history of second-degree consanguinity, and father of the child has nystagmus and microphthalmos. On examination he had a vision of OU – c/us/m with OD : CTC : +0.50 DS −0.75 DC 180 AXIS OS : CTC : −1.00 DS −0.50 DC 180 AXIS correction. He had a left exotropia with the right face turn and OU pendular nystagmus with normal pigmented iris. Systemic examination is unremarkable. Dilated fundus examination revealed normal optic nerve heads and absent foveal reflex with the vessels passing over the area of incipient fovea (Figures 1 and 2).

OCT was carried out, which showed the absence of foveal depression with the persistence of the inner retinal layers. The diagnosis of isolated primary foveal hypoplasia was made, and the patient was prescribed glasses and asked for a regular follow up (Figures 3 and 4).

Discussion
There were no signs of regressed ROP, myopia, cataract, corneal pannus, ocular albinism or IP as has been reported previously1–4 in association with the foveal hypoplasia in our patient.

The role of OCT in the diagnosis of this case was also obvious. There was preservation of the inner retinal layers in the foveae of both the eyes as demonstrated by the OCT.
This report illustrates the importance of careful examination of the foveal area in patients with unexplained visual loss with nystagmus that does not fit into the other well-known dystrophies. The report also highlights the role of OCT in the diagnosis of this condition, by the demonstration of the preservation of the inner retinal layers in the fovea.

References

**Figure 4:** OCT of the left eye which showed the absence of foveal depression with the persistence of the inner retinal layers. Foveal thickness OS 282 µm.