

Case Report Ophthalmology

Isolated foveal hypoplasia - A rare case report

Gitumoni Sharma¹, Sagarmoy Purkayastha²

¹Vitreous-Retina, ROP, Uveitis Services, ²Cataract and Glaucoma Services ASG Eye Care, Paltanbazar, Guwahati, Assam, India.

***Corresponding author:**

Sagarmoy Purkayastha,
Cataract and Glaucoma
Services, ASG Eye Care, Paltan
Bazaar, Guwahati, Assam, India.

drspurkayastha@rediffmail.com

Received: 07 September 2025

Accepted: 27 September 2025

Published: 10 October 2025

DOI

10.25259/TEE_7_2025

Quick Response Code:



ABSTRACT

Objectives: The objective is to present the case of a male child with isolated foveal hypoplasia.

Material and Methods: A 3.5-month-old male child with an inability of visual fixation was referred by a pediatrician for ophthalmological examination. There was nystagmus. Dilated fundus examination revealed a dull foveal reflex. Electroretinography and VEP (visual evoked potential) were abnormal. However, no familial association was established. On follow-up at 4 years, he was detected for moderate visual impairment with high hyperopia. Fundus examination showed the macular and foveal reflexes to be absent, and the retinal capillaries were abnormally close to the presumed foveolar area. After 9 years of follow-up, fundus reflexes appear as before. OCT (optical coherence tomography) showed the absence of foveolar depression and reduced retinal thickness, diagnosing it to be isolated foveal hypoplasia. The child is otherwise healthy and performing well in school, similar to other children of his age.

Results: Following a complete ophthalmological examination and a long follow-up, the clinical suspicion of isolated foveal hypoplasia in this otherwise healthy patient was established.

Conclusion: With this report, we want to highlight the roles of electrophysiology, fundus photography, and OCT in the diagnosis of a rare condition like isolated foveal hypoplasia.

Keywords: Albinism, Foveal hypoplasia, Microphthalmos, Nystagmus, Optical coherence tomography

INTRODUCTION

The central retina consists of a specialized pit structure called the fovea. When the foveal pit does not develop fully, it is called foveal hypoplasia and is associated with poor vision. The literature contains only a few reports of foveal hypoplasia as an isolated anatomical finding. Noval *et al.*, in a cross-sectional study with 286 healthy children, found that up to 3% of children had an anatomically underdeveloped foveal pit or fovea plana bilaterally on OCT.¹

The foveal avascular zone results from capillary closure after birth, is independent of vision, and the vessels occasionally seen traversing this zone represent a persistence of the fetal vasculature. The above description might also apply to foveal hypoplasia, but confirmatory, embryological, and histological evidence is lacking. Also, no definite hereditary pattern is found.

Recently, it has been explained that autosomal dominant isolated foveal hypoplasia (FVH1) is a rare condition of foveal hypoplasia (FH) that lacks any other ocular manifestations. FVH1 is associated with hypomorphic mutations in the *PAX6* gene that encodes a sequence-specific

This is an open-access article distributed under the terms of the Creative Commons Attribution-Non Commercial-Share Alike 4.0 License, which allows others to remix, transform, and build upon the work non-commercially, as long as the author is credited and the new creations are licensed under the identical terms.

©2025 Published by Scientific Scholar on behalf of The Eastern Eye.

DNA-binding transcription factor for morphogenesis and evolution of the eye.²

They reported on 17 patients with *PAX6* mutations associated with FVH1 or FH with aniridia and corneal opacities. Patients with three mutations, p.V78E, p.V83F, and p.R128H, in the C-terminal subdomain (CTS) of the paired domain consistently have severe FH. The findings demonstrated that mutations associated with FVH1 underline a functional divergence between DNA-binding ability and transcriptional activity. They concluded that a wide range of mutations in the *PAX6* gene is not limited to the CTS region and is responsible for FVH1.

Isolated foveal hypoplasia has been reported in association with albinism, aniridia, microphthalmos, and occasionally achromatopsia,³ which are its differentials as well.

Fundus autofluorescence and fundus fluorescence angiography are less sensitive diagnostic tools.⁴ Optical coherence tomography (OCT), as a noninvasive and quick method, is helpful in the diagnosis of foveal hypoplasia.⁵ OCT reveals absent foveal depression in eyes with maintenance of all the inner retinal layers and outer plexiform layer in the typical foveal pit location. On multimodal colour imaging (MCI), hyporeflexivity corresponding to normal foveal depression is characteristically absent on blue (BR) and green reflectance (GR) images. The infrared reflectance (IR) image is normal. In the presence of nystagmus, obtaining optimum quality and precise OCT scan images may be difficult. Enface images on MCI confirm the absent of foveal depression even in the presence of gross nystagmus, and it can provide another way to identify foveal hypoplasia, which complements the existing gold standard of OCT imaging.⁶

CASE REPORT

At 3.5 months, a male baby was referred by his pediatrician for not developing visual fixation. The parents complained of the baby's head turning mostly to the right side and that he does not follow much light or sound. There is a history of preterm birth at 32 weeks with a normal delivery. Birth weight was 2.5 kg. Under direct illumination, anterior segment examination was normal in both eyes. Dilated fundus examination showed dull foveal reflex and mild disc elevation in both eyes. Visual evoked response in electroretinography (ERG) showed reduced rods and cones response (cones > rods), suspected of cone-rod dystrophy in both eyes [Figure 1]. Ganzfeld flash VEP gave an impression of both eyes' diffuse RPE atrophy and pendular nystagmus [Figure 2]. Parents were informed about his condition. However, no familial association could be established.

The patient came back after 4 years, with parents complaining of difficulty seeing the blackboard. Best corrected visual acuity was 6/36 N10 with +1.50DS/+1.00DC × 90 in the right eye and

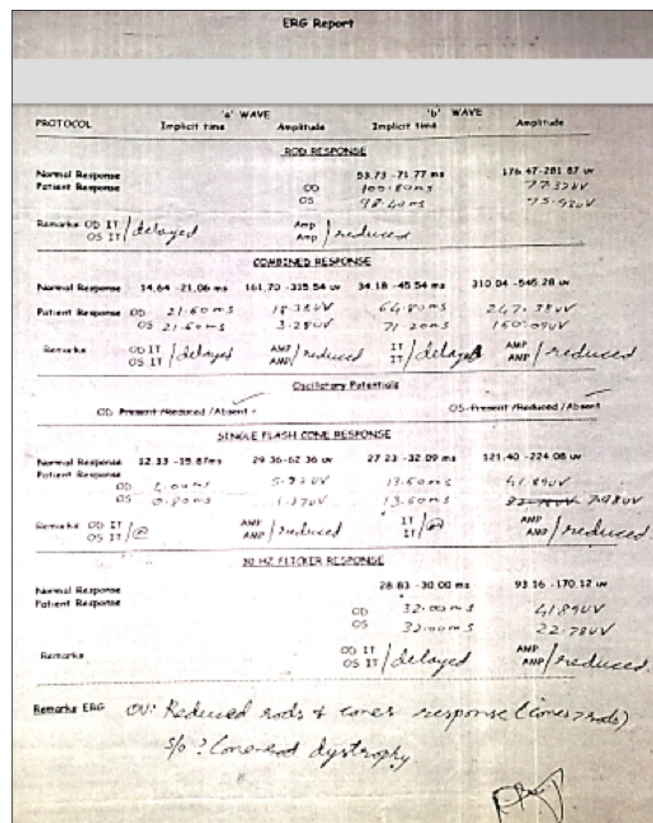


Figure 1: ERG report shows reduced rod-cone response (cones > rods), consistent with cone-rod dystrophy. ERG: Electroretinogram.

6/24 N10 with +2.50DS/+1.00DC × 90 in the left eye after wet retinoscopy. Nystagmus was persistent. The anterior segment was normal in each eye. However, the fundus showed the macular and foveal reflexes to be absent, and the retinal capillaries were abnormally close to the presumed foveolar area [Figure 3].

He was advised to wear glasses. After nine years since his first presentation, the child is maintaining the same vision. Nystagmus is persistent. Anterior segment is normal. Fundus is shown in Figure 3. OCT done shows absence of foveolar depression and the same retinal thickness in all the scans, as is shown in Figures 4 and 5, confirming isolated foveal hypoplasia. The child is otherwise healthy and performing well in school, similar to the other children of his age.

DISCUSSION

Foveal hypoplasia has been described as a lack of foveal depression with continuity of all neurosensory retinal layers in the presumed foveal area.⁷⁻⁹ It is well known in aniridia, albinism, microphthalmus, achromatopsia, and retinopathy of prematurity, which are its differential diagnoses.

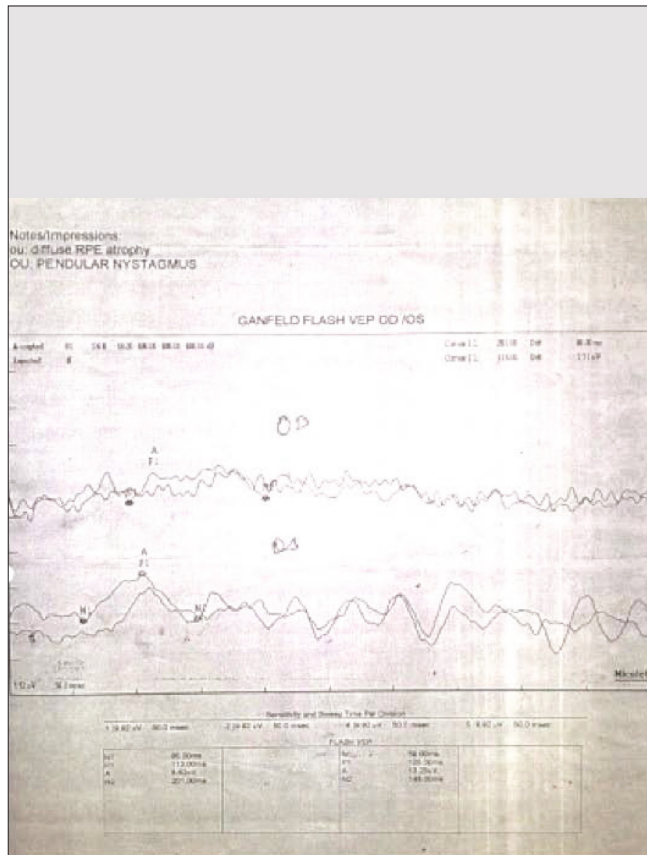


Figure 2: Ganzfeld VEP shows bilateral diffuse RPE atrophy and pendular nystagmus. VEP: Visual evoked potential, RPE: Retinal pigment epithelial.



Figure 3: Fundus photograph of the posterior pole in isolated foveal hypoplasia; (a) Right eye, (b) Left eye.

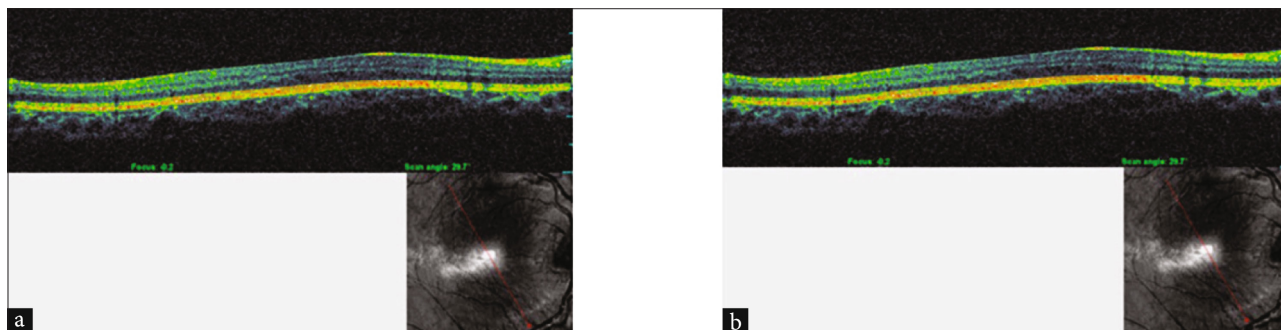


Figure 4: OCT (a) Right eye, (b) Confirming foveal hypoplasia. OCT: Optical coherence tomography.

Aniridia is a congenital eye disorder featured by the partial or complete absence of the iris, where foveal hypoplasia is sometimes present, often due to mutations in the *PAX6* gene.

Albinism is a group of genetic conditions causing reduced pigmentation in the skin, hair, and eyes, along with foveal hypoplasia, which may be a common feature, along with nystagmus and strabismus, making it a crucial part of the differential diagnosis.

Microphthalmus is a condition in which one or both eyes are abnormally small and may also present with foveal hypoplasia due to shared genetic or developmental causes.

Achromatopsia is again a rare, inherited eye disorder that affects colour vision and also presents with foveal hypoplasia, reduced visual acuity, and photophobia. Unlike typical foveal hypoplasia, achromatopsia can also involve photoreceptor degeneration.

There is one more condition called incontinentia pigmenti, which is again a rare genetic disorder that affects the skin, teeth, hair, nails, and nervous system, and can also involve ocular anomalies, including foveal hypoplasia.

Mostly discussed these days is the retinopathy of prematurity, which affects premature infants and can lead to various retinal vascular abnormalities, including foveal hypoplasia in some cases.

Foveal hypoplasia as an isolated finding is rare, only about 0.4%.⁹ In recent years, OCT has been described as a quick and confirmatory tool to confirm the suspected diagnosis. Rarely, isolated foveal hypoplasia causes infantile nystagmus¹⁰ as in this case. Affected patients have no other signs of albinism and no chiasmal misrouting. This condition should be considered in the child who presents with infantile nystagmus with a negative family history. Optical coherence tomography and multifocal ERG are useful in confirming the diagnosis, as is shown in the case. Mutations in the *PAX6* homeobox gene have been identified in this condition.^{2,10} Individuals with infantile nystagmus also demonstrate a significant association between more severe foveal hypoplasia and increasing hyperopia¹¹ as is present in this case.

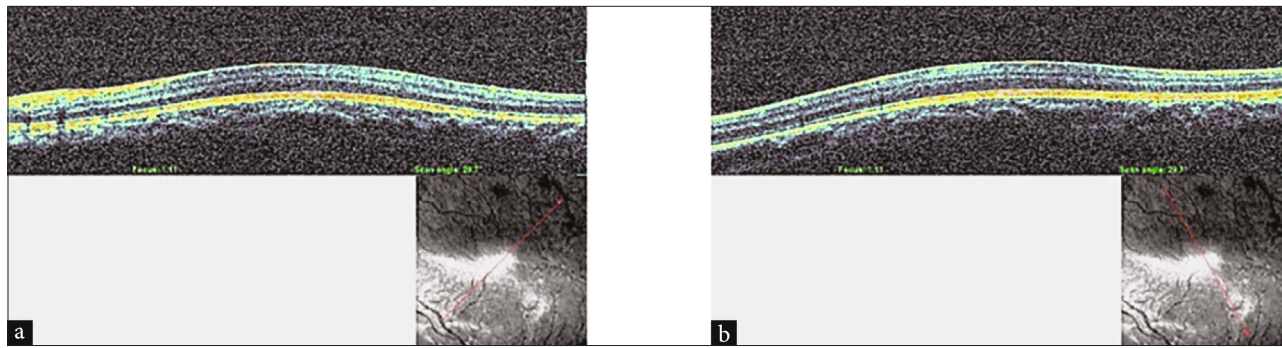


Figure 5: OCT (a) Left eye, (b) Confirming foveal hypoplasia. OCT: Optical coherence tomography.

In this case, after having the report of ERG, cone-rod dystrophy in both eyes is suspected, and the Ganzfeld flash VEP gave an impression of diffuse RPE atrophy and pendular nystagmus in both eyes. However, multifocal ERG, which was not available, would have demonstrated reduced P1 amplitudes in the central rings 1, 2, and 3.¹²

CONCLUSION

Clinically, the diagnosis of isolated foveal hypoplasia can be made by disproportionate visual loss in children, with or without systemic conditions of albinism. They maintain a workable vision for the long term and are capable of doing day-to-day activities.

This case, which was referred from a pediatrician for non-fixation of gaze at 3.5 months, when the diagnosis in particular becomes a real challenge. Rare cases like this can be diagnosed only if follow-up is done properly with the investigations as and when required. Long years of follow-up are sometimes required.

Acknowledgement: The authors would like to thank the Retina Institute of Karnataka, Bengaluru, for providing all scientific support in preparing the case report.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship: Nil.

Conflicts of interest: There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation: The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Noval S, Freedman SE, Asrani S, El-Dairi MA. Incidence of fovea plana in normal children. *J AAPOS* 2014;18:471–5.

2. Matsushita I, Izumi H, Ueno S, Hayashi T, Fujinami K, Tsunoda K, *et al.* Functional characteristics of diverse PAX6 mutations associated with isolated foveal hypoplasia. *Genes (Basel)* 2023;14:1483.
3. Oliver MD, Dotan SA, Chemke J, Abraham FA. Isolated foveal hypoplasia. *Br J Ophthalmol* 1987;71:926–30.
4. Mota A, Fonseca S, Carneiro A, Magalhães A, Brandão E, Falcão-Reis F. Isolated foveal hypoplasia: tomographic, angiographic and autofluorescence patterns. *Case Rep Ophthalmol Med* 2012;2012:864958.
5. Holmström G, Eriksson U, Hellgren K, Larsson E. Optical coherence tomography is helpful in the diagnosis of foveal hypoplasia. *Acta Ophthalmologica* 2010;88:439–42.
6. Venkatesh R, Jain K, Yadav NK. Multicolour imaging in isolated foveal hypoplasia. *Indian J Ophthalmol* 2019;68:191–2.
7. Querques G, Bux AV, Iaculli C, Noci ND. Isolated foveal hypoplasia. *Retina* 2008;28:1552–3.
8. Vincent A, Kemmanu V, Shetty R, Anandula V, Madhavarao B, Shetty B. Variable expressivity of ocular associations of foveal hypoplasia in a family. *Eye* 2009;23:1735–9.
9. Issa PC, Foerl M, Helb HM, Scholl HPN, Holz FG. Multimodal fundus imaging in foveal hypoplasia: Combined scanning laser ophthalmoscope imaging and spectral-domain optical coherence tomography. *Arch Ophthalmol* 2008;126:1463–5.
10. Brodsky MC. *Paediatric neuro-ophthalmology*. 2nd edition. New York: Springer; 2010. p. 400.
11. Healey N, McLoone E, Mahon G, Jackson AJ, Saunders KJ, McClelland JF. Investigating the relationship between foveal morphology and refractive error in a population with infantile nystagmus syndrome. *Invest Ophthalmol Vis Sci* 2013;54:2934–9.
12. Sanghi G, Gupta R, Brar A. Photo essay. *Indian J Ophthalmol* 2023;3:569–71.

How to cite this article: Sharma G, Purkayastha S. Isolated foveal hypoplasia - A rare case report. *East Eye*. 2025;1:40–43. doi: 10.25259/TEE_7_2025