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STARGARDT DISEASE DIAGNOSED WITH SPECTRAL-DOMAIN OCT AND PERIMETRY IN THE ABSENCE OF FUNDUS AUTOFLUORESCENCE AND FLUORESCEIN ANGIOGRAPHY: A CASE REPORT

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ABSTRACT

Purpose: To report a case of Stargardt disease diagnosed on the basis of clinical phenotype, spectral-domain optical coherence tomography (SD-OCT), and automated perimetry, in the absence of fundus fluorescein angiography (FFA) and fundus autofluorescence (FAF). **Methods:** Detailed ophthalmic evaluation, multimodal imaging, and functional tests were performed. **Results:** A 36-year-old male presented with progressive bilateral central vision loss. Fundus examination revealed characteristic yellow–white flecks and macular atrophy. OCT demonstrated ellipsoid zone disruption, outer nuclear layer thinning, and hyperreflective deposits at the level of the retinal pigment epithelium. Automated perimetry showed dense central scotomas with preserved peripheral fields. **Conclusion:** In settings where FAF and FFA are not available, a combination of clinical findings, OCT, and perimetry can provide sufficient evidence to establish a diagnosis of Stargardt disease.

INTRODUCTION

Stargardt disease or Fundus Fluvimaculatus is the most common inherited juvenile macular dystrophy[1], usually presenting in the first two decades of life with progressive bilateral central vision loss. It is caused primarily by mutations in the ABCA4 gene located in Chromosome 1p22.1, leading to accumulation of lipofuscin within the retinal pigment epithelium (RPE).[2] Classic diagnostic imaging relies on fundus fluorescein angiography (FFA), demonstrating the "dark choroid" sign, and fundus autofluorescence (FAF), which highlights lipofuscin deposition.[3]

However, in resource-limited settings, these investigations may not be readily available. Advances in optical coherence tomography (OCT) and visual function testing, including perimetry, now provide valuable diagnostic alternatives.[3] A case of Stargardt disease diagnosed using OCT and perimetry in the absence of FFA and FAF, highlighting their clinical utility is reported.

Presenting History

A 36-year-old male presented with complaints of gradually progressive, bilateral central vision loss since childhood. There was no significant family history of ocular disorders or systemic disease. Blurring of vision increased in last 3 months with increased sensitivity to bright light and difficulty in finer works like reading, recognising faces or writing. He developed difficulty in recognising between red and green colours from the last 1 month in both eyes. It was associated with grey spots in vision. Adjustment difficulties noted when moved to dark places from areas of bright light.

Ocular Examination

Visual acuity was [4/60 OD, 4/60 OS]. Slit-lamp examination of the anterior segment was unremarkable, and intraocular pressure were normal in both eyes. Fundus examination by indirect ophthalmoscope revealed multiple yellow–white pisciform flecks scattered at the posterior pole, with macular atrophy showing a beaten-bronze appearance. Peripheral retina and optic discs were normal bilaterally.

Colour vision testing revealed red–green discrimination defects with a score of 2/21 plates in Ishihara pseudoisochromatic chart.

Investigations

In the absence of fundus fluorescein angiography and fundus autofluorescence, spectral-domain optical coherence tomography (SD-OCT) was employed as the principal imaging modality. OCT demonstrated disruption and loss of continuity of the ellipsoid zone at the fovea, with thinning of the outer nuclear layer. Multiple hyperreflective deposits were visible at the level of the retinal pigment epithelium (RPE), corresponding to the clinically observed flecks. Central macular thinning was also noted, consistent with progressive macular atrophy.

Standard automated perimetry (Humphrey 30-2) revealed dense central scotoma with relative preservation of the peripheral field.

USG B-Scan was also done which was found to be normal bilaterally.

Provisional Diagnosis

The clinical phenotype, together with characteristic OCT and perimetry findings, supported the diagnosis of Stargardt disease. Genetic confirmation was not feasible due to resource constraints.

Management

Patient was advised to continue with antioxidant tablets twice daily after meal along with Vitamin A tablet supplementations. BCVA was done up to 6/60 (OD) and 6/60 (OS). Patient was also referred to the department of Physical Medicine and Rehabilitation (PMR) for vocational rehabilitation.

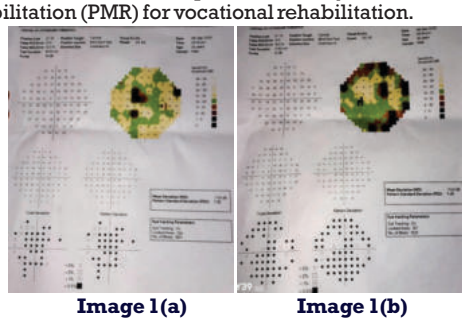

Image 1(a) **Image 1(b)**

Image 1: Perimetry Reports of the Patient 1(a) Right Eye & 1 (b) Left Eye

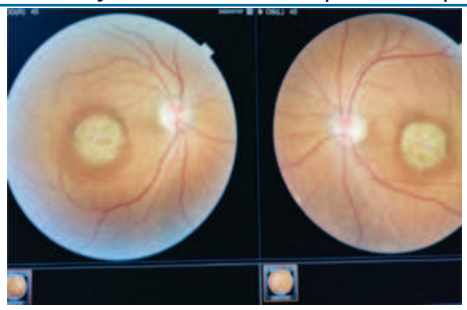

Image 2: OCT Fundus Photo of Right and Left Eye

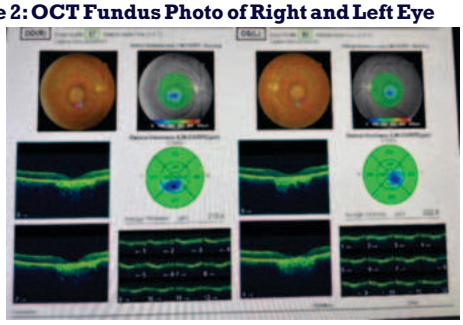

Image 3: OCT Macula Photo of Right and Left Eyes

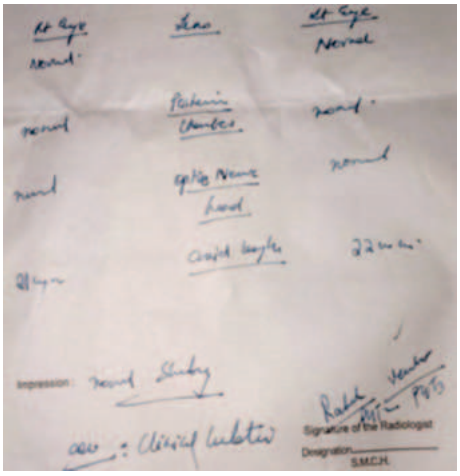

Image 4: USG B-Scan Report of Right and Left Eyes

DISCUSSION

Stargardt disease typically presents with central vision loss, characteristic fundus flecks, and macular atrophy. FFA and FAF are considered diagnostic standards; however, absence of these modalities can make diagnosis challenging.

OCT is a reliable tool to demonstrate hallmark structural changes: disruption of the ellipsoid zone, thinning of the outer retinal layers, hyperreflective RPE deposits, and macular atrophy.[3] In this case, OCT findings correlated well with fundus appearance.

Functional tests add diagnostic strength. Perimetry typically reveals central or paracentral scotomas with preserved peripheral vision, as was observed in this case.[4] Colour vision defects and decreased visual acuity further support macular dysfunction.

Differential diagnoses include cone dystrophy, Best vitelliform macular dystrophy, and pattern dystrophy. However, the presence of characteristic flecks, OCT findings, and perimetric scotomas favours Stargardt disease.

This case highlights the importance of multimodal evaluation in resource-limited settings. Even in the absence of FFA and FAF, a confident diagnosis can be achieved using OCT and perimetry.

CONCLUSION

Stargardt disease can be effectively diagnosed through a combination of clinical features, OCT imaging, and perimetry when standard angiographic and autofluorescence techniques are unavailable. This case emphasizes the diagnostic utility of OCT and visual function testing in resource-limited clinical practice.

Consent

Informed consent was obtained from the patient for sharing his reports and other clinical information understanding that due efforts will be made to conceal his identity but anonymity cannot be guaranteed.

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REFERENCES

1. Altschwager P, Ambrosio L, Swanson EA, Moskowitz A, Fulton AB. Juvenile Macular Degenerations. Semin Pediatr Neurol. 2017 May;24(2):104-109. doi: 10.1016/j.spen.2017.05.005
2. Cremers, F. P. M., Lee, W., Collin, R. W. J., & Allikmets, R. (2020). Clinical spectrum, genetic complexity and therapeutic approaches for retinal disease caused by ABCA4 mutations. Progress in retinal and eye research, 79, 100861. doi: 10.1016/j.preteyeres.2020.100861
3. Tanna P, Strauss RW, Fujinami K, Michaelides M. Stargardt disease: clinical features, molecular genetics, animal models and therapeutic options. Br J Ophthalmol. 2017 Jan;101(1):25-30. doi: 10.1136/bjophthalmol-2016-308823
4. Messias, A., Reinhard, J., Velasco e Cruz, A. A., Dietz, K., MacKeben, M., & Trauzettel-Klosinski, S. (2007). Eccentric fixation in Stargardt's disease assessed by Tübingen perimetry. Investigative ophthalmology & visual science, 48(12), 5815–5822. https://doi.org/10.1167/iovs.06-0367