Title: Handheld Optical Coherence Tomography in Young Infant with Albinism and Fovea Plana

Running title: Infant with Fovea Plana

Authors: Sohaib R. Rufai (BMBS, MRes)[1,2], Helena Lee (PhD, FRCOphth)[3], Irene Gottlob (MD, FRCOphth)[1]*

Affiliations:

1. The University of Leicester Ulverscroft Eye Unit, Leicester Royal Infirmary, Robert Kilpatrick Clinical Sciences Building, PO Box 65, Leicester LE2 7LX, United Kingdom.
2. Clinical and Academic Department of Ophthalmology, Great Ormond Street Hospital NHS Foundation Trust, London WC1N 3JH, United Kingdom.
3. Clinical and Experimental Sciences, Faculty of Medicine, University of Southampton, Southampton SO16 6YD, United Kingdom.

*Corresponding author:

Professor Irene Gottlob, MD, FRCOphth

Address: The University of Leicester Ulverscroft Eye Unit, Leicester Royal Infirmary, Robert Kilpatrick Clinical Sciences Building, PO Box 65, Leicester LE2 7LX, United Kingdom.

E-mail: ig15@leicester.ac.uk

Tel: +44 (0)116 2523268

Fax: +44 (0)116 2231996

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Abstract

We present handheld optical coherence tomography (OCT) diagnosis of grade 4 foveal hypoplasia (fovea plana) in a 28-day-old infant with albinism. Grade 4 foveal hypoplasia is characterised by the absence of the foveal pit, absence of outer segment lengthening and absence of outer nuclear layer widening. Binocular VA at 58 months follow-up was 1.2 logMAR. We describe our handheld OCT acquisition protocol and compare the morphological features with a healthy, age-matched control subject.

Article text:

Here, we present optical coherence tomography (OCT) imaging obtained from a 28-day-old Caucasian male patient with albinism, referred to our clinic for workup of infantile nystagmus. On examination, the patient had pendular nystagmus, transillumination defects and cutaneous hypopigmentation. The patient was unable to cooperate with preferential looking testing.

Figure 1A displays successful handheld OCT acquisition in this patient, without dilation or sedation, demonstrating Grade 4 foveal hypoplasia (fovea plana).[1] Grade 4 foveal hypoplasia is characterised by the absence of the foveal pit, absence of outer segment lengthening and absence of outer nuclear layer widening. OCT was performed using a handheld device (ENVISU C class 2300 [Leica Microsystems, Wetzlar, Germany]; <4-µm axial resolution). The acquisition protocol used a 10×10-mm scanning window. The 3-dimensional raster scan program for both scan sequences comprised 100 B-scans and 500 A-scans per B-scan line. The acquisition time was short (1.9 seconds) to facilitate successful image acquisition with minimal disruption of quality, thereby avoiding measurement bias. **Figure 1B** displays a normal fovea from a healthy, age-matched, Caucasian male control for comparison. On latest follow-up at 58 months old, the patient's binocular VA was 1.2 logMAR.

Albinism is associated with nystagmus and foveal hypoplasia.[1-3] Typical foveal hypoplasia can be graded on a scale of 1-4, based on morphological characteristics seen on optical coherence tomography (OCT) imaging, while atypical foveal hypoplasia is seen exclusively in achromatopsia.[1] In our unit's recent study, we found that foveal hypoplasia grading was the best predictor of future VA compared to preferential looking testing, foveal developmental index (the ratio of inner retinal layers v.s. total foveal thickness), outer segment length and photoreceptor length.[1] Preferential looking testing was only successful in 68.1% of patients, compared to a 90% success rate for handheld OCT.[1] Of note, our study found that infants with grade 4 foveal hypoplasia had mean predicted VA of 1.0 logMAR at school age.[1] In this illustrative case, the patient's final VA fell within 2 lines of this figure.

Predicting future VA using OCT can help parents in planning adjustments to optimise their child's development and educational attainment.[4] From the clinician's perspective, the high degree of foveal hypoplasia in this case likely accounts for the limitation in VA. However, if VA was worse than predicted according to foveal hypoplasia grading, then suspicion should be raised for other co-existing pathology limiting the child's VA. This could include amblyopia, uncorrected refractive error, anterior segment conditions, retinal dystrophy or neurological disorders.

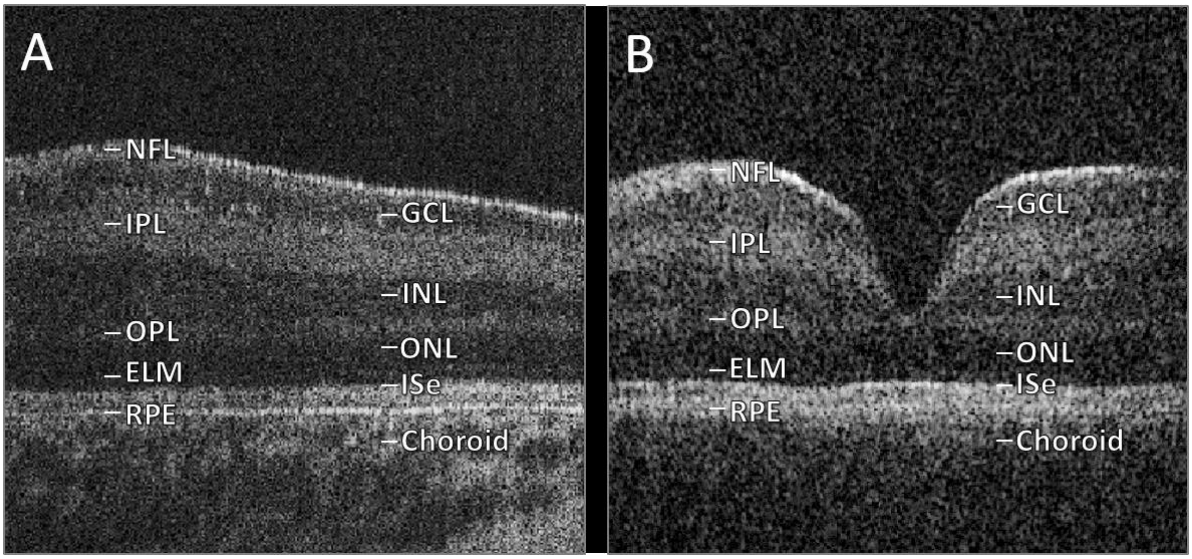

Figure 1: OCT images for 28-day-old with fovea plana (1A) and 24-day-old control subject (1B). Key: NFL = nerve fibre layer; GCL = ganglion cell layer; I/OPL = inner/outer plexiform layer; I/ONL = inner/outer nuclear layer; ELM = external limiting membrane; ISe = inner segment ellipsoid; RPE = retinal pigment epithelium.

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