

Spectral domain optical coherence tomography and microperimetry in foveal hypoplasia

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A case of foveal hypoplasia associated with ocular albinism with anatomic and functional changes by various techniques using spectral domain optical coherence tomography (SD-OCT), microperimeter and confocal scanning laser ophthalmoscope is described. This case highlights the importance of microperimeter in detecting the functional abnormalities of vision and SD-OCT in identifying the retinal laminar abnormalities in foveal hypoplasia.

Key words: Foveal hypoplasia, microperimetry, spectral domain optical coherence tomography

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Albinism is a relatively rare (1 in 20,000 people) mutation of one or more genes. It is categorized into oculocutaneous and ocular albinism, based on the involvement of both skin and eyes versus only eyes respectively. It is a defect in the generation of the melanin pigment in oculocutaneous albinism, and decreased number of melanosomes with normal amount of melanin in ocular albinism. Foveal hypoplasia is the characteristic feature of albinism often associated with aniridia, microphthalmus, nystagmus and achromatopsia.^[1]

We present a case of foveal hypoplasia, in which spectral domain optical coherence tomography (SD-OCT), microperimetry and fundus autofluorescence (FAF) were performed to explain the structural and functional characteristics.

Case Report

We examined a 33-year-old male with progressive painless

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decrease in vision and nystagmus since childhood and no history of night blindness. His best corrected visual acuity was 20/80 with 0.00 diopter sphere (D Sph) / -4.00 diopter cylinder (D Cyl) X 10° in the right eye and 20/60 with 0.00 D Sph / -3.50 D Cyl X 170° in the left eye respectively. Color vision was normal on examination with Ishihara plates. A positive history of similar abnormality was obtained in his sibling (elder brother had similar abnormality). There was no history of consanguinity. Slit-lamp examination was normal and iris did not show any transillumination defect. Intraocular pressure using applanation tonometry was 15 mm of Hg in both eyes. Fundus examination showed macular vascularization, macular hypoplasia and hypoplastic optic nerve head with diffuse retinal pigment epithelial atrophy and prominent choroidal vessels in both eyes. Patient was clinically diagnosed to have foveal hypoplasia. A genetic analysis revealed an X-linked recessive inheritance pattern.

SD-OCT (Copernicus, Italy) was done in both eyes using asterisk scan protocol (7 mm length scan, 6 B-scans with 2743 A-scans per B-scan). SD-OCT was recorded with the eyes in the null zone, to overcome nystagmus. The scan location was assured by positioning the asterisk scan acquisition pattern over the foveal area and this location was assured as the scan passes through the fovea in the fundus image obtained in SD-OCT. The scan revealed absence of foveal dip, preservation of the inner retinal layers with thickened fovea, 270 microns in the right eye and 245 microns in the left eye. The qualitative and quantitative characteristics of the retina were similar in macular and peripheral areas (2° and 7° respectively). The small elevation of the inner segment/ outer segment (IS/OS) junction was absent in the fovea which is otherwise present in normal eyes.

Microperimetry (MP1, Nidek Technologies, Padova, Italy) using 33 stimuli in the central 20° of the macula and 4-2 threshold strategy revealed reduction of retinal sensitivity in the central retina. Mean retinal sensitivity was 7.2 dB in the right eye and 14.2 dB in the left eye. Relative scotoma was present in both eyes at the foveal area. Fixation was relatively unstable in both eyes, with abnormal fixation location (central fixation in the right eye and eccentric fixation in the left eye). Average eye movements during the microperimetry were 1.27°/sec in the right eye and 1.49°/sec in the left eye. Figs. 1 and 2 describe the color fundus photography, microperimetry and SD-OCT findings in our case. Fundus Autofluorescence (FAF) imaging with confocal scanning laser ophthalmoscope (Heidelberg Retina Angiograph, HRA II, Heidelberg Engineering, Heidelberg, Germany) did not show the typical foveal darkening in both eyes [Fig. 3].

Discussion

Foveal hypoplasia in oculocutaneous albinism is commonly associated with ocular features like decreased visual acuity, nystagmus, photophobia, strabismus, iris transillumination, macular transparency and neuronal abnormality.^[1,2] Ours is a unique case of foveal hypoplasia associated with ocular albinism. High refractive errors are more common in patients with albinism.^[3] In our case both eyes had high astigmatism with reasonably good visual acuity in contrast to previously reported studies.^[1,4,5] Our case is Grade 3 (grading system proposed by Seo *et al.*,^[1]) in view of mild foveal hyporeflectivity,

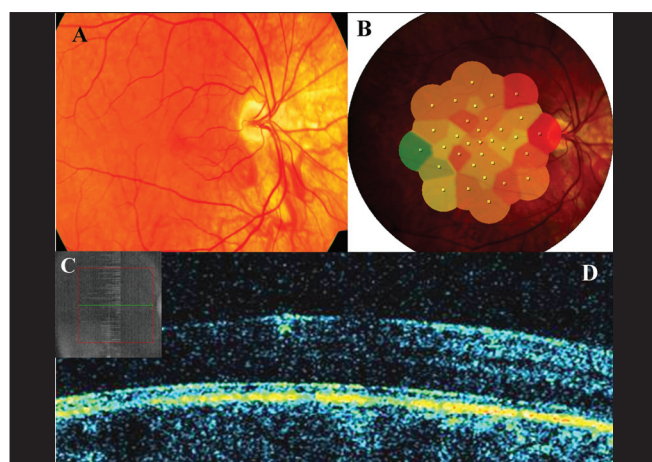


Figure 1: (A) Color fundus photograph of the right eye reveals macular vascularization and disc hypoplasia. (B) Microperimetry shows reduced retinal sensitivity in the central macular area. (C) Fundus-reconstructed image from SD-OCT showing the location of the scan. (D) SD-OCT image shows absence of foveal depression and persistence of inner retinal layers at the fovea

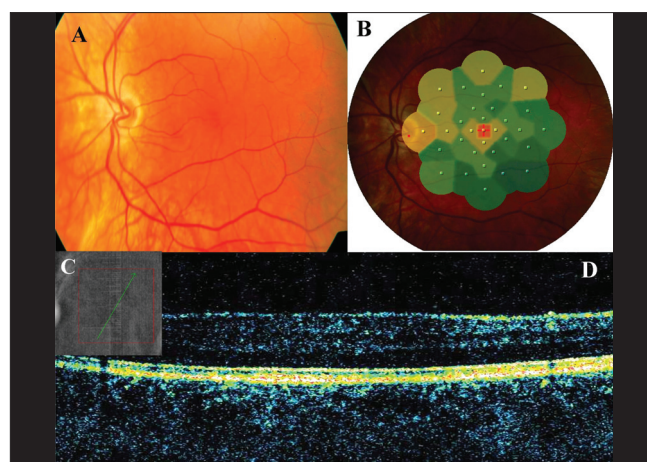


Figure 2: (A) Color fundus photograph of the left eye reveals macular vascularization and disc hypoplasia. (B) Microperimetry shows reduced retinal sensitivity in the central macular area. (C) Fundus-reconstructed image from SD-OCT showing the location of scan. (D) SD-OCT image shows absence of foveal depression and persistence of inner retinal layers at the fovea

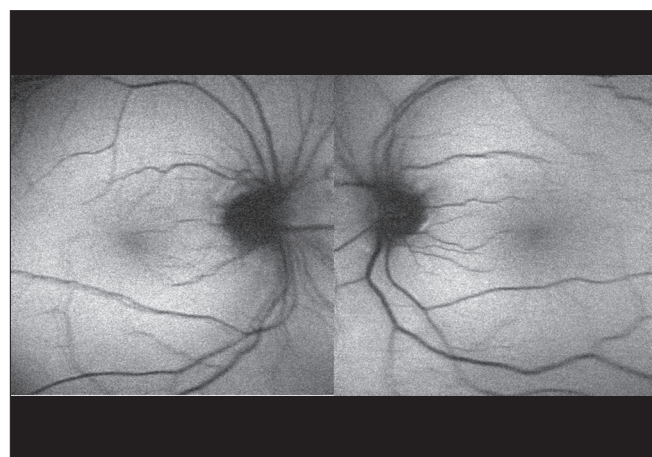


Figure 3: Fundus autofluorescence of right eye and left eye shows reduced autofluorescence at the macula

<2 choroidal transillumination, absence of tram-tract sign and foveal depression.

SD-OCT showed absence of foveal depression, as reported in previous studies using time domain OCT.^[1,4-6] The foveal thickness in this case was higher due to the presence of inner retinal layers in the foveal area. This can be due to the absence of the Chievitz layer which has an important role in the foveal maturity.^[2,7] In the normal fovea, the photoreceptor layer appears as a distinct hyper-reflective layer with high-resolution OCT due to the difference in the optical properties of the photoreceptor segments,^[8] and the distance between the retinal pigment epithelium and the IS/OS junction of the photoreceptors increases significantly in the foveal region, consistent with the well-known increase in the length of the outer cone segments in this region.^[9] In case of foveal hypoplasia this layer is not very distinct as the central cones are spaced apart.^[1] Probably this explains low visual acuity in

these patients. In our case we found persistent IS/OS junction of the photoreceptor layer in the foveal area in both the eyes but the central IS/OS junctional elevation was absent. This could be due to no difference in the size of the cones present in the foveal and peripheral retina.

Microperimetry showed a dense scotoma in the central macular area. The retinal sensitivity was less in the central macular area compared to the peripheral area. Abnormal cones in the central fovea cause low vision which affect the emmetropization process and can lead to nystagmus. In our case the patient had jerky nystagmus and the average eye movement measured with microperimetry was relatively higher compared to the normal age-matched control (0.18°/sec, unpublished data). The fixation of this patient was relatively unstable in both the eyes due to nystagmus. FAF of the central macula was reduced which may be due to the amount of macular pigment present.^[10]

In the literature only one case of isolated foveal hypoplasia has been reported. Our case report shows foveal hypoplasia associated with ocular albinism in which SD-OCT and microperimetry were used for the first time. Our study shows the importance of the microperimeter in assessing the functional vision.

References

1. Seo JH, Yu YS, Kim JH, Chung HK, Heo JW, Kim SJ. Correlation of visual acuity with foveal hypoplasia grading by optical coherence tomography in albinism. *Ophthalmology* 2007;114:1547-51.
2. Querques G, Bux AV, Iaculli C, Delle Noci N. Isolated foveal hypoplasia. *Int Ophthalmol* 2009;29:271-4.
3. Wildsoet CF, Oswald PJ, Clark S. Albinism: Its implications for refractive development. *Invest Ophthalmol Vis Sci* 2000;41:1-7
4. Meyer CH, Lapolice DJ, Freedman SF. Foveal hypoplasia in oculocutaneous albinism demonstrated by optical coherence tomography. *Am J Ophthalmol* 2002;133:409-10.
5. McGuire DE, Weinreb RN, Goldbaum MH. Foveal hypoplasia demonstrated in vivo with optical coherence tomography. *Am J*

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- Ophthalmol 2003;135:112-4.
6. Recchia FM, Carvalho-Recchia CA, Trese MT. Optical coherence tomography in the diagnosis of foveal hypoplasia. Arch Ophthalmol 2002;120:1587-8.
 7. Vedantham V. Isolated foveal hypoplasia detected by optical coherence tomography. Indian J Ophthalmol 2005;53:276-7.
 8. Drexler W, Morgner U, Ghanta RK, Kartner FX, Schuman JS, Fujimoto JG. Ultrahigh-resolution ophthalmic optical coherence tomography. Nat Med 2001;7:502-7.
 9. Drexler W, Sattmann H, Hermann B, Ko TH, Stur M, Unterhuber A, *et al.* Enhanced visualization of macular pathology with the use of Ultrahigh-resolution optical coherence tomography. Arch Ophthalmol 2003;121:695-706.
 10. Charbel Issa P, Foerl M, Helb HM, Scholl HP, 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.
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