Photo Essay

Multimodal imaging in dominant cystoid macular dystrophy

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Dominant cystoid macular dystrophy (DCMD) is a rare inherited retinal disorder. It primarily affects the macula. It is a unique retinal dystrophy because the appearance of cystic spaces in the macula heralds its onset with the rest of the retina being essentially normal.[1] Herein, we describe multimodal imaging findings in a case of DCMD.

Case Report

A 30-year-old male patient presented to us with gradual diminution of vision in both eyes for 1 year. There was no history of night blindness and no history of niacin intake in the past. Family history was not significant. Best-corrected visual acuity was 20/30; N6 in both eyes. Anterior segment was normal with clear lens. There was no evidence of inflammation in the anterior chamber. Vitreous was clear. In both eyes, discs were healthy, retinal vessel caliber was normal, and pars plana was clear. Both eyes had cystoid macular edema at the posterior pole [Fig. 1]. Multicolor imaging showed a greenish hue with central orange reflectance suggestive of gross cystoid macular edema [Fig. 2]. Both short wave autofluorescence and near infra-red autofluorescence (NIR-AF) showed central multispot hyperautofluorescence and an area of hyperautofluorescence involving the posterior pole and extending up to the arcades [Fig. 3]. NIR-AF showed an additional area of hyperautofluorescence, nasal to the disc in both eyes [Fig. 4]. Spectral domain optical coherence tomography showed cystoid spaces involving all the retinal layers in both eyes [Fig. 5]. Fluorescein angiography (FA) showed faint petaloid leakage at the fovea in both eyes [Fig. 6]. The patient refused electrophysiological tests. Based on history, clinical examination and imaging findings a diagnosis of DCMD was made.

Discussion

DCMD is a rare disease. The disease usually has its onset in the third decade of life. As the disease progresses the cystic spaces diminish giving rise to chorioretinal atrophy with subsequent loss in vision.[2] It has been suggested that pathophysiologically, this disease is caused by both abnormal muller cells as well as retinal pigment epithelial (RPE) dysfunction.[1] The current report highlights the multimodal imaging signatures of this entity. Multicolor imaging helps in picking up the cystoid abnormality much efficiently as compared to the conventional color fundus photograph. Fundus autofluorescence imaging, especially NIR-AF unmasks a larger area of RPE disturbance not picked up either on clinical examination or FA; thus, giving credence to the RPE dysfunction hypothesis. NIR-AF will be extremely useful to prognosticate and in follow-up of these cases, replacing invasive investigations such as fundus fluorescein angiography.

Multicolor imaging has the potential to completely replace color fundus photography in the future because both morphological and functional definition of the retina is rendered into a single composite image with the added advantages that all required image acquisitions can be performed with a single machine and in a nondilated pupil. It may also serve as a valuable noninvasive modality to determine the natural course or response to treatment based on regression of the area of greenish-hue representative of macular edema in these patients.

Electrophysiological tests and genetic testing are necessary addendum to the above-mentioned imaging modalities for conclusive diagnosis.

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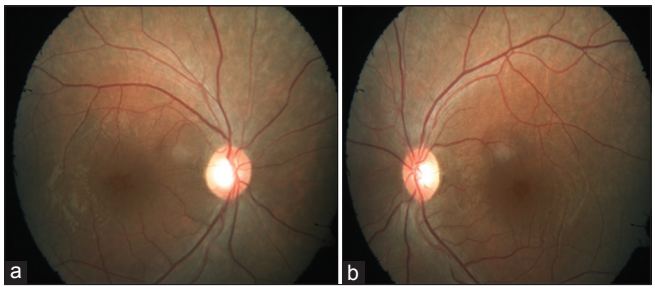

Figure 1: Color fundus photograph of right (a) and left (b) eye showing cystoid macular edema at posterior pole

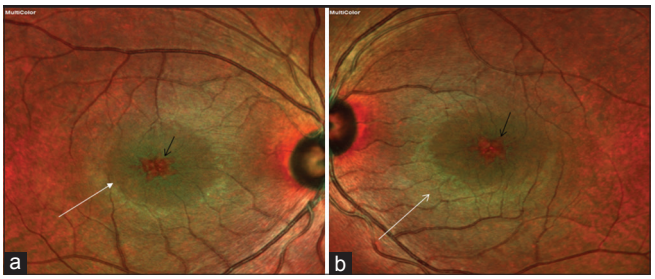

Figure 2: Multicolor image of right (a) and left (b) eye showing greenish hue at posterior pole suggestive of retina thickening (white arrows) with central orange hyper reflectance at fovea (black arrows) suggestive of a cyst

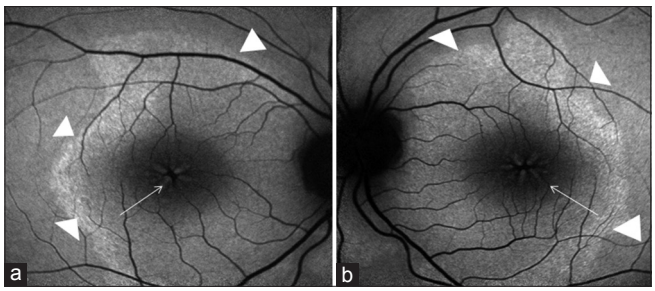

Figure 3: Short wavelength autofluroscence of right (a) and left (b) eye showing distinct multispot hyperautofluorescence at the fovea (white arrows) with surrounding larger area of hyperautofluorescence involving the posterior pole extending beyond the arcades (white arrowheads)

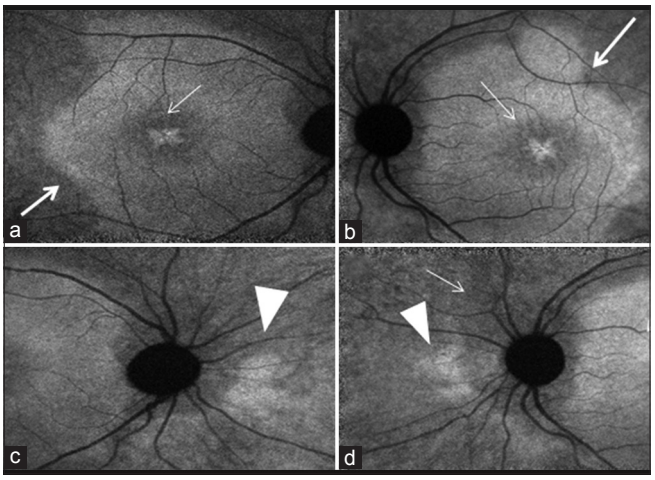

Figure 4: Near infra-red autofluroscence of right (a) and left (b) eye showing distinct multispot hyperautofluorescence at the fovea (thin white arrows) with surrounding larger area of hyperautofluorescence involving the posterior pole extending beyond the arcades (thick arrow). Hyperautofluorescent areas noted nasal to disc in both eyes (arrowhead c and d)

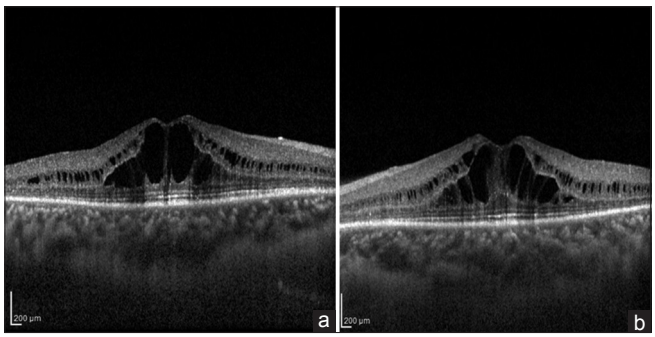

Figure 5: Spectral domain optical coherence tomography line scan through the fovea of both right (a) and left (b) eye showing cystoid spaces involving all retinal layers

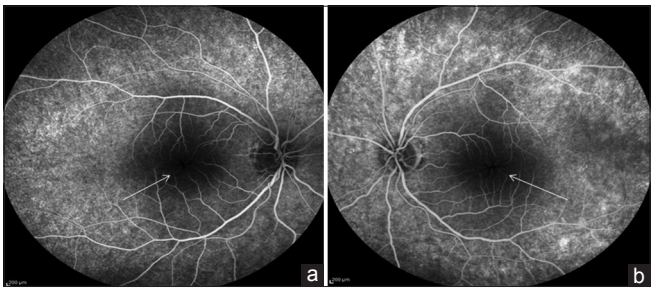

Figure 6: Fundus fluorescein angiography midphase of right (a) and left (b) eye showing very faint petaloid hyperfluorescence (white arrows) at the fovea

Conclusion

The present report describes the multimodal imaging including multicolor findings in DCMD and provides unique insight into its possible pathogenesis.

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Nil.

Conflicts of interest

There are no conflicts of interest.

References

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2. Ganesh A, Stroh E, Manayath GJ, Al-Zuhaibi S, Levin AV. Macular cysts in retinal dystrophy. Curr Opin Ophthalmol 2011;22:332-9.