Multimodal imaging in a case of choroideremia

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Choroideremia (CHM) is a rare X-linked disorder that causes progressive degeneration of retina, retinal pigment epithelium (RPE) and choroid.[1,2] Multicolor imaging (MCI) is a noninvasive retinal imaging modality.[3] We report hithertho unreported MCI characteristics of CHM.

A 30-year-old male presented with complaints of bilateral night blindness. Best corrected visual acuity was 20/30 bilaterally. Fundus examination showed bilateral chorioretinal atrophy (CRA) and areas of RPE disruption with sparing of central macula [Fig. 1a and b]. Electroretinogram showed nonrecordable cone and rod responses. MCI (Spectralis; Heidelberg) [Fig. 1c and d] highlighted the residual RPE tissue at macula and the surrounding CRA much better than color fundus photography. Blue autofluorescence (BAF; 488 nm; Spectralis; Heidelberg) showed generalized decreased autofluorescence with residual areas of autofluorescence in the macular area representing RPE disease [Fig. 2a and b]. The residual RPE tissue was well visualized in infrared reflectance (IR) [Fig. 3a and c] as compared to green [Fig. 4a and c] and blue reflectances [Fig. 4b and d]. The spectral domain optical coherence tomography showed retinal thinning and choriocapillary atrophy [Fig. 3b and d]; however, genetic testing was not available.

CHM usually starts from the periphery and a central retinal island is usually preserved until late in the disease course. Patients usually have good vision until degeneration involves fovea. Amount of viable retina in CHM can be assessed reliably by BAF. BAF usually shows bilateral, symmetric, midperipheral zones of hypoautofluorescence due to RPE atrophy with preserved area of central stellate autofluorescence.[4] In the present case, the amount of viable

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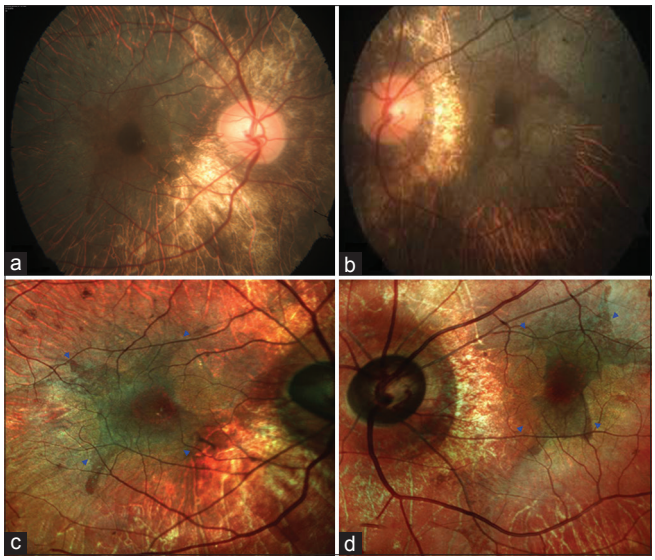

Figure 1: Color fundus photograph (CFP) (a and b) showed bilateral chorioretinal atrophy (CRA) and areas of retinal pigment epithelium (RPE) disruption with sparing of the central macula. Multicolor imaging (c and d) highlighted the residual RPE tissue at macula (blue arrow heads) and surrounding CRA much better than CFP

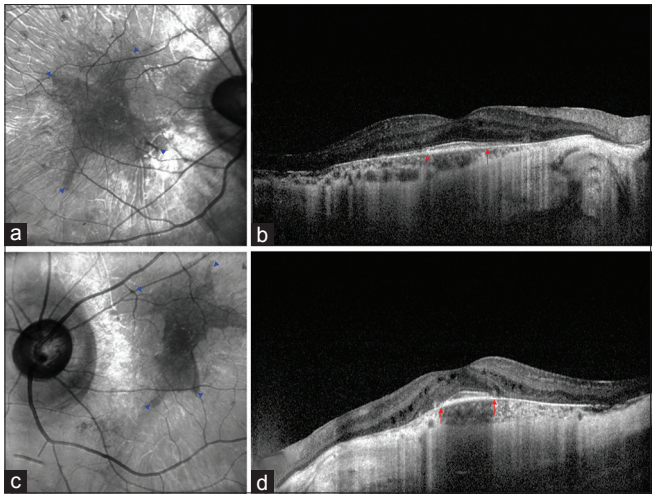

Figure 3: Area of residual RPE tissue (blue arrow heads) was well visualized in infrared reflectance images (IR) (a and c). Spectral domain optical coherence tomography showed retinal thinning and choriocapillary atrophy sparing central macula (red arrows) (b and d)

retina is picked up clearly by MCI. This area was best seen in IR channel because it visualizes structures at the level of outer retina, RPE and choroid. This report highlights the utility of MCI in evaluating viable retina in CHM. MCI may be used to document disease progression in CHM.

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Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

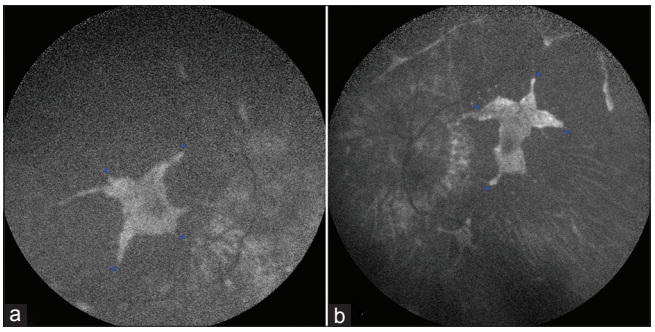

Figure 2: Blue fundus autofluorescence images in both eyes (a and b) showed generalized decreased autofluorescence with residual areas of autofluorescence in the macular area (blue arrow heads)

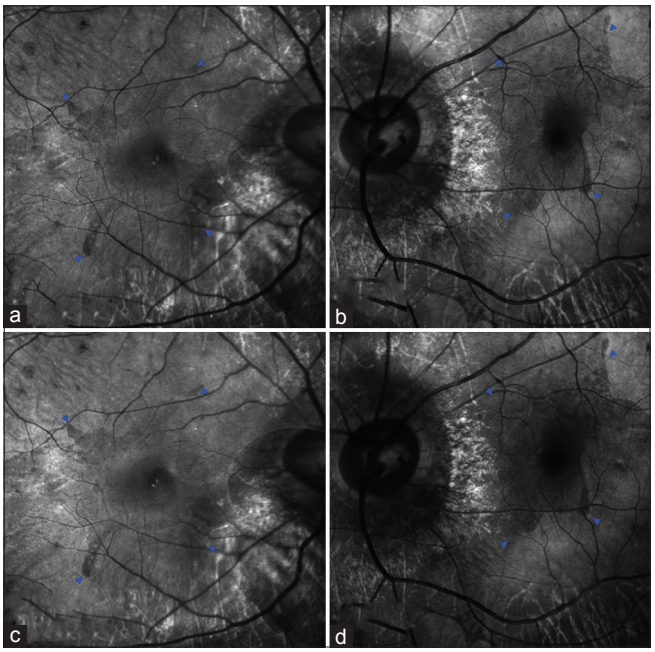

Figure 4: Green reflectance images (a and c) and blue reflectance images (b and d) of both eyes showed area of residual RPE tissue (blue arrow heads) faintly as compared to IR

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Conflicts of interest

There are no conflicts of interest.

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