Utility of multimodal imaging in the detection and follow-up of retinal astrocytic hamartomas in a young girl with tuberous sclerosis complex

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Key words: Blue and green reflectance, multimodal imaging, optical coherence tomography, retinal astrocytic hamartoma

Retinal astrocytic hamartomas (RAH) are one of the major diagnostic criteria for tuberous sclerosis complex (TSC) and is conventionally detected by ophthalmoscopy. We describe the utility of multimodal imaging (MMI) with spectral domain optical coherence tomography (SDOCT) and multicolor imaging (MCI) in the detection of hamartomas which may be missed clinically, especially in pediatric patients who may not be able to cooperate for detailed fundus examination. We also present some novel imaging findings which helped to understand the evolution of these lesions.

A 5-year-old girl with tuberous sclerosis complex (TSC) was subjected to retinal evaluation and MMI. Fundus examination showed subtle elevation superotemporal to the disc in the right eye (RE) and a circular greyish lesion overlying superotemporal arcade in the left eye (LE) [Fig. 1a, b]. Corresponding OCT showed dome-shaped elevation of the nerve fiber layer (NFL), confirming the lesions as RAH [Fig. 1c, d]. MCI with its component blue and green reflectance (BR, GR) well delineated these superficial lesions with uniform hyporeflectivity. MCI supported by OCT revealed additional lesions in both eyes which were not visible clinically. Right eye was found to have a well-defined superficial lesion along the inferotemporal arcade with minimal elevation of the NFL and intact outer retina on OCT [Fig. 2a]. LE had two small deep lesions, one superiorly and another temporally, with minimal or no elevation of the NFL, outer retinal collapse and ellipsoid zone loss on OCT [Fig. 2b, c]. On BR and GR, superficial lesion in the RE had hyporeflectance [Fig. 3a, b], whereas the deep lesions in the LE were hyperreflective with central hyporeflectance [Fig. 4a]. At review six months later, the temporal lesion in the LE showed an additional hyporeflective outer rim [Fig. 4b]. OCT was unchanged at follow-up.

Discussion

OCT features of RAH are well described and improves the detection of these lesions.[1–4] A majority of RAH remain stable, but can rarely regress spontaneously.[5] MCI with its component BR and GR is an additional tool to detect and monitor these lesions. We report novel findings on MMI in RAH and correlate lesion characteristics with the evolution of the lesion. We assumed superficial RAH with elevation of NFL on OCT to be active lesions. The deep RAH with predominant changes in the outer retina and progressive changes on BR and GR may be regressing lesions and may require MMI for detection. While BR and GR showed progressive changes on follow-up, OCT features remained stable, thereby indicating that MCI may be more sensitive to monitor the course of RAH in TSC.

Although RAH indicates tumor-like growth in inner retinal layers which usually remain constant, the use of BR and GR with OCT could aid in diagnosis and follow-up of RAH.

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

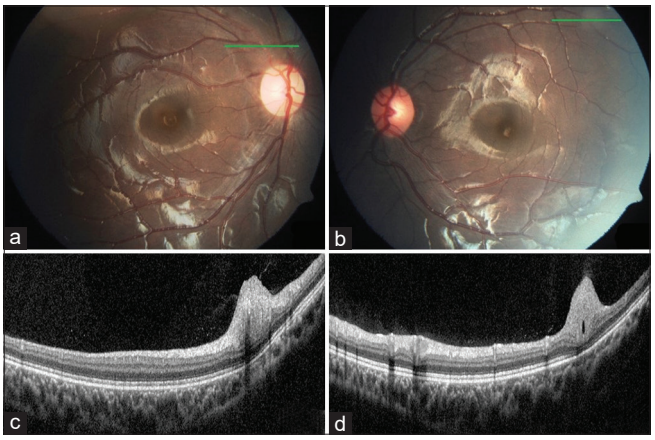

Figure 1: Colour fundus photo (CP) and optical coherence tomography (OCT) of both eyes: CP (a and b) showed retinal astrocytic hamartomas superior to the disc in the right eye (RE) and over the superotemporal arcade in the left eye (LE). OCT through the lesions showed dome-shaped elevation of the nerve fiber layer with back-shadowing in RE (c) and a hyporeflective cavitation in LE (d)

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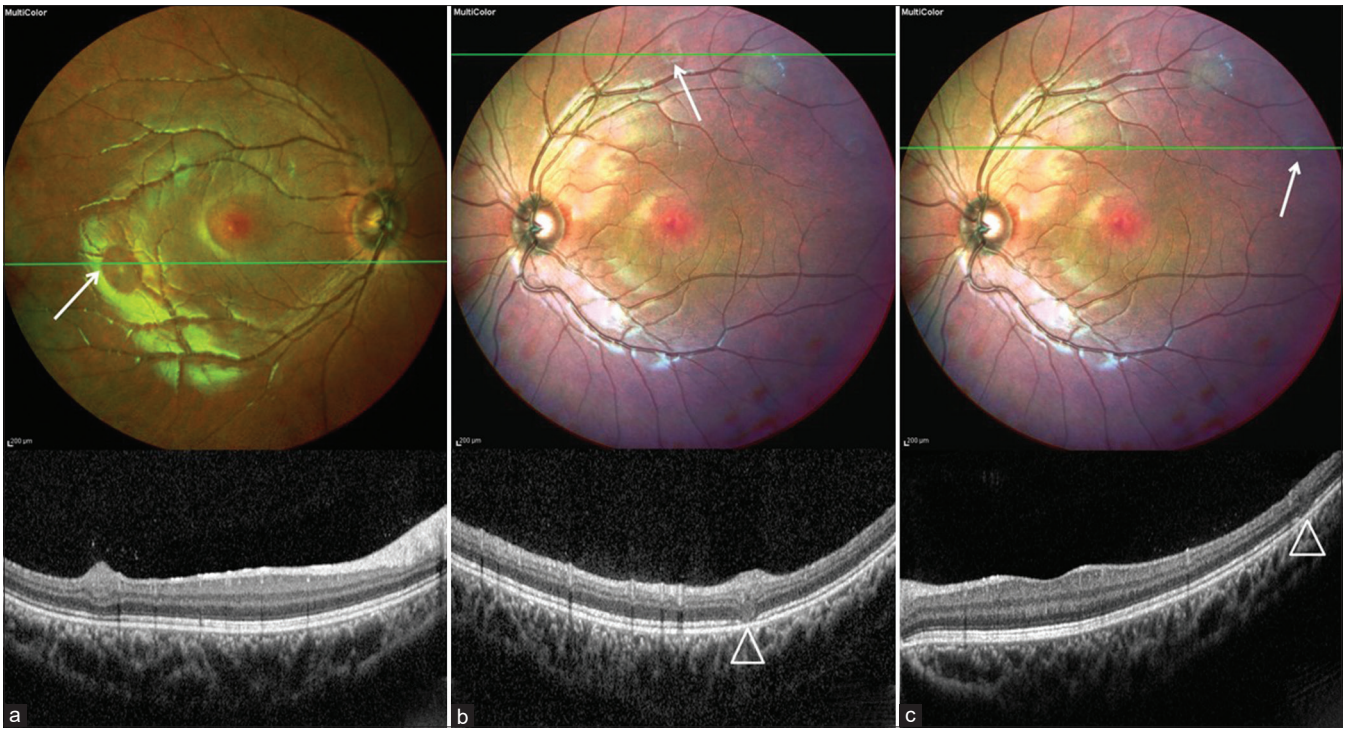

Figure 2: Multicolor imaging (MCI) with (OCT) of both eyes: MCI demonstrated additional lesions in both eyes (arrow). On OCT, superficial lesion in the RE (a) showed elevation of the nerve fiber layer with intact outer retina, whereas deeper lesions in the LE (b and c) showed predominant changes in the outer retina with outer retinal collapse and ellipsoid zone loss (arrowheads)

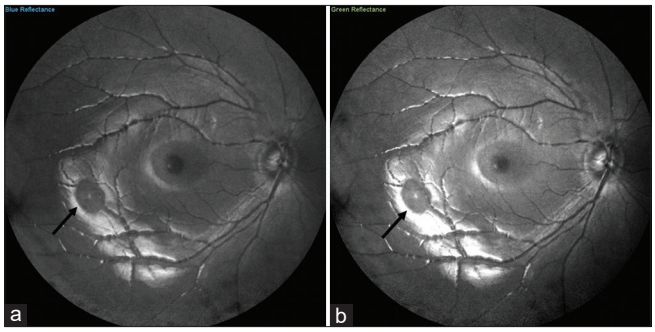

Figure 3: Blue (a) and green (b) reflectance (BR, GR) of the right eye: A well-delineated elevated lesion with uniform hyporeflectivity was noted over the temporal vessels (arrow). The confocal high-resolution BR and GR imaging with superior contrast enabled improved detection of the inner retinal lesion which was missed on ophthalmoscopy and colour fundus photography

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.

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Conflicts of interest

There are no conflicts of interest.

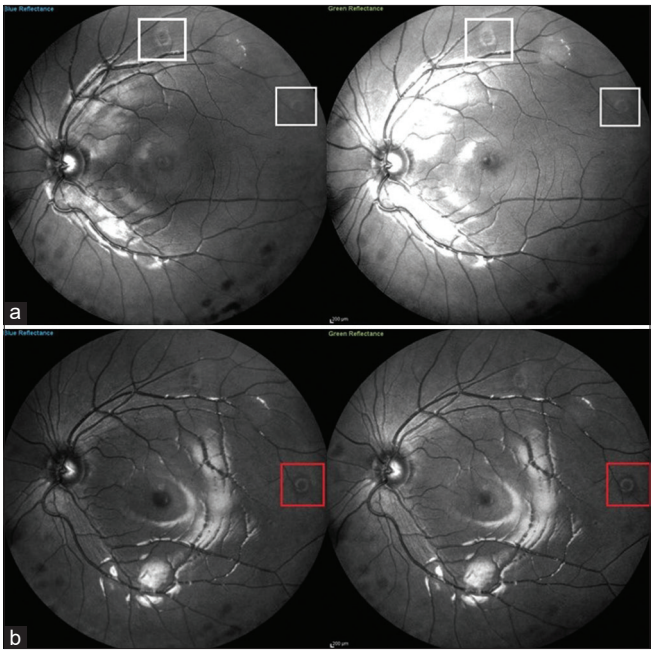

Figure 4: Blue and green reflectance (BR and GR) of the left eye: At presentation, BR and GR of the deep lesions showed hyperreflectivity with central hyporeflectance (a, white squares). At six months follow-up, the superior lesion remained stable while the temporal lesion showed an additional hyporeflective outer rim in a characteristic Bull's eye configuration (b, red squares)

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