Subfoveal congenital hypertrophy of retinal pigment epithelium

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DESCRIPTION

A 25-year-old man presented with slight blurring of vision in the right eye (RE), noted 3 years ago. There was no positive systemic history. His best corrected visual acuity was 6/9 in RE and 6/6 in the left eye (LE) at presentation. Anterior segment evaluation was unremarkable in both eyes with normal pupil light reflexes. Fundus evaluation revealed a flat circular lesion beneath the fovea, and was measured to be one-third of the size of the optic nerve head. It was a densely pigmented lesion, black in colour, with a surrounding depigmented whitish halo (figure 1). The LE was within normal range on clinical examination. Spectral domain optical coherence tomography (SD-OCT) of RE revealed the lesion to be irregular but flat with hyper-reflectivity at the level of the retinal pigment epithelium (RPE). Discontinuity of the ellipsoid zone was noted to be overlying some parts of the lesion. These findings were also accompanied by foci of intraretinal migration of the pigment epithelium within the outer nuclear layer and a subretinal cleft (figure 2). The central macular thickness was measured to be 252μm. No consistent scotoma was detected on repeated charting with Amsler's grid. After careful literature review of OCT findings of hyperpigmented lesions, this lesion was labelled as subfoveal congenital hypertrophy of RPE (CHRPE) and was advised regular follow-up.

CHRPE is usually present in the peripheral fundus with nearly all demonstrating a perilesional halo.[1] It may or may not have lacunae within it. Macular CHRPE is very rare, and subfoveal rarer still.[1 2] It is believed to represent less than 1% of all the cases of CHRPE.[2] Therefore, vision loss due to

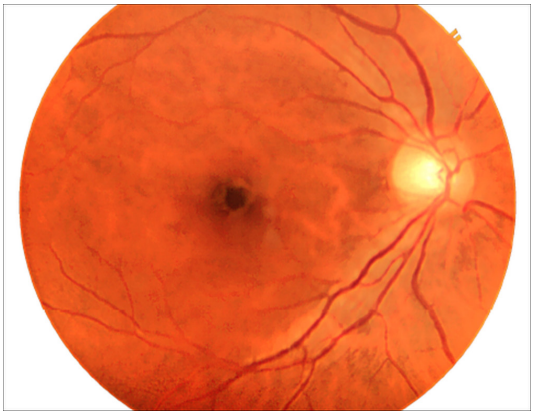

Figure 1 Fundus photograph of the right eye showing the well-defined flat subfoveal lesion. The lesion is densely pigmented and has a regular halo around it.

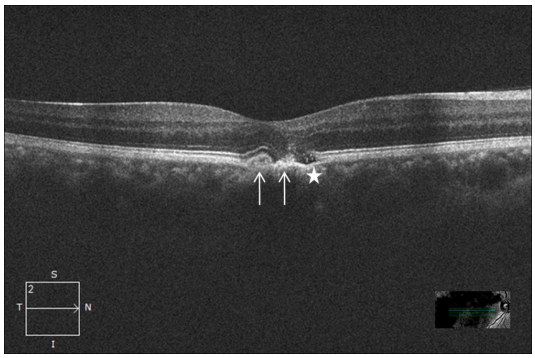

Figure 2 Spectral domain optical coherence tomography horizontal line scan through the lesion (seen in inset infrared image) showing hyper-reflectivity at the level of the retinal pigment epithelium (RPE) along with a thickened RPE layer (white arrows). Intraretinal migration of RPE, deficiency of ellipsoid layer and external limiting membrane, and a subretinal cleft are noticeable (star).

CHRPE is rare. A retrospective case series on OCT findings found these lesions to be flat with thickened and irregular RPE, with overlying neurosensory retina to be affected until the outer nuclear layer (67%) in majority of the cases. Intraretinal findings included hyper-reflective foci along with subretinal clefts in the regions where outer retinal layers were disturbed.[1] The authors describe subretinal fluid to be an absence of layers wherein the tissue appears to be retracted in a shallow manner. Photoreceptor loss has been repeatedly shown to be an important feature of CHRPE.[1] All these features were present in our case too (figure 2).

Differential diagnosis of dark pigmented macular lesions includes haemorrhagic degenerations like polypoidal choroidal vasculopathy which is seen in older age groups. It is progressive in nature with raised lesions and choroidal changes with commonly accompanying subretinal fluid. Further, choroidal tumours like melanoma may also simulate these

Learning points

- Congenital hypertrophy of retinal pigment epithelium may rarely involve the subfoveal region and result in minor visual disturbance. The signs, however, are out of proportion to the symptoms.
- Knowledge of optical coherence tomography features helps in diagnosis and decreases the requirement of expensive and invasive investigations.

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lesions and vice versa. However, OCT features of these tumours have shown subretinal fluid accompanying dome-shaped lesions with shaggy photoreceptors.[3] Thus apart from the clinical phenotype, knowledge of the OCT features aids in diagnosing uncommon presentations and obviates expensive and invasive investigations. Though the lesion looked ominous in this patient (figure 1), the visual loss was minor and stationary.

Contributors HS worked on the patient, performed imaging and did patient counselling. BT diagnosed the patient. BT, VS and HS wrote the manuscript. BT and SVA critically revised the manuscript.

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