• Letter to the Editor •

A case of congenital simple hamartoma of the retinal pigment epithelium combined with fascicular nerve fibre layer defects

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Dear Editor,

We observed a patient with congenital simple hamartoma of the retinal pigment epithelium (CSHRPE) using multimodal imaging, which helped to differentiate this disease from other retinal pigment epithelial diseases. CSHRPE is a rare benign lesion. It is typically characterized by an isolated, round and dark black lesion located in the center of the macula, which was first described by Laqua[1]. It is currently considered a congenital benign lesion, but its pathogenesis remains unclear[2-3].

Ethical Approval The patient signed the informed consent form, but since it was a retrospective case report that we did not make an ethics application.

A 52-year-old female presented with a 1-year history of physical examination findings of fundopathy in the left eye that was not associated with vision loss. The patient's previous history included 6mo of hypertension and 2y of diabetes mellitus; both were well controlled. The patient had no history of surgery or trauma. Ophthalmological examination revealed a visual acuity of 1.0 in both eyes, a noncontact intraocular pressure of 19 mm Hg and no significant abnormality in the anterior segment of the eye. At presentation, ophthalmoscopic examination revealed a dark gray–black lesion at the upper vascular arch. The lesion was characterized by a well-demarcated border approximately 1/5 papillary diameter in size. The upper edge of the lesion crossed the branches of the superior temporal retinal artery and was combined with a bundle of nerve fibre layer loss (Figure 1). In addition, there were scattered drusen on the temporal side of the fovea. No obvious abnormalities were detected in the right eye. Structural optical coherence tomography (OCT) revealed a hyperreflective lesion and underlying structures with shadowing effects (Figure 2). OCT angiography (OCTA) revealed rich blood flow signals. Clustered vasculature was visible in the en face images (Figure 2). In the early stage, fundus fluorescein angiography (FFA) of the left eye showed a well-defined hypofluorescent lesion, which gradually became vascular clustered structures with mild leakage of fluorescein in the late stage (Figure 2).

CSHRPE is a benign lesion with supplying vessels. It was first reported by Shields *et al*[4] under this name. The characteristic features of CSHRPE include a single gray–black lesion mostly less than 1.5 mm in diameter. It is currently thought to be congenital but is usually detected late because it is asymptomatic. At an even more forward time, Gass[3] classified it as a congenital retinal pigment epithelial anomaly in the form of a focal, nodular, dark black lesion and predicted the course of the disease, which usually manifests itself as involvement of the entire retina and spills over onto the inner surface of the retina in an umbilical fashion, typically ranging from 1/2 to 1 papillary diameter in size; frequently occurring in the macular region[5]; independent of neuroepithelial, retinal pigment epithelium, and choroidal changes; and no exudates or hemorrhages. For such a single pigment lesion, it needs to be differentiated from solitary congenital hypertrophy of retinal pigment epithelium[6], combined hamartoma of the retina and retinal pigment epithelium[7], choroidal nevi[8] and retinal pigment epithelial adenoma[9]. Although the majority of reported patients with CSHRPE have isolated lesions, a rare case of multifocal CSHRPE has recently been reported. A Spanish female has two hyperpigmented lesions in the center of the macula in the fundus[10].

With the progress and application of multimodal imaging technology, the level of understanding of the disease is

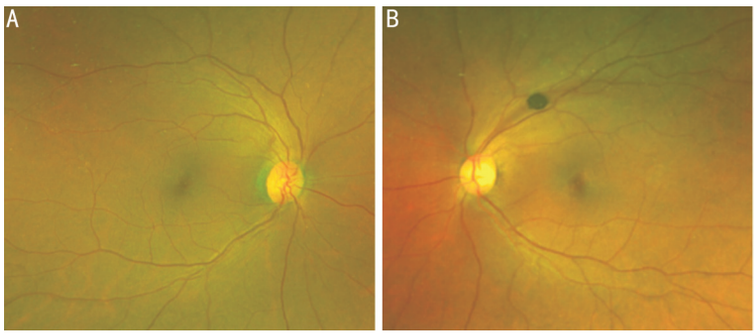

Figure 1 Optos fundus photography showing a normal fundus in the patient's right eye (A) and CSHRPE at the superior temporal vascular arch in the left eye, with a fascicular nerve fibre layer defect and no peripheral haemorrhage or exudation (B) CSHRPE: Congenital simple hamartoma of the retinal pigment epithelium.

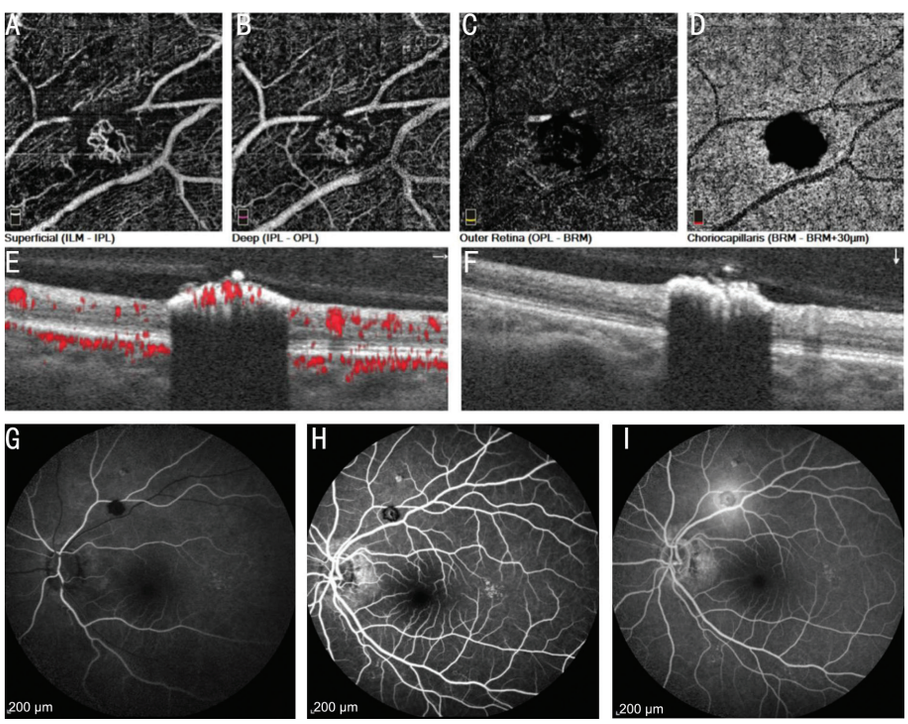

Figure 2 OCTA showed lesions with distinct clustered blood vessels in both superficial and deep retinal layers (A, B), exhibiting low signal in the outer retinal and choroidal capillary layers (C, D), and containing abundant blood flow signal inside the lesion (E), and structural OCT showed lesions with irregular hyperreflection, steep edges, and shadowing of the underlying structures (F); FFA is seen as a hypofluorescent lesion in the early stages, with vascular cluster-like structures appearing internally in the middle and late stages, accompanied by a mild hyperfluorescent halo in the late stages (G-I). OCTA: Optical coherence tomography angiography; OCT: Optical coherence tomography; FFA: Fundus fluorescein angiography.

gradually improving[11]. In the 5 patients reported by Shields[4], no notable vascular structures were detected *via* FFA due to pigment occlusion, and there was no significant fluorescein leakage at late stages. In contrast, the patient in our case had visible clumped vascular structures on FFA. A slight leakage may be related to the location of the lesion in a more superficial layer of the retina. In addition, changes in the permeability of the vascular wall accompanying the prolonged course of the disease cannot be excluded. On structural OCT, it is a highly reflective structure in the superficial retinal layer with an irregular surface accompanied by steep edges that obscure all the structures below, sometimesthe Humpty Dumpty sign is presented[12]. FFA signals are prone to blockage by pigmented tissue or fibrosis in the mass. OCTA can overcome these limitations and clearly reveal the vascular system of the tumor. Its signal is unmasked by the pigmentation of the lesion. Vascular structures are present among the layers[13-14] and can be further distinguished from other retinal pigment epithelial tumors[15].

Due to the low prevalence of the disease, only case reports are currently available in the literature. However, the CSHRPE usually remains stable. Ito and Ohji[16] reported a patient with 10y of follow-up with a lesion located in the central macula. The patient suffered a mild decrease in visual acuity during

the first 3y, which remained stable throughout the following 7y. However, rare complications can inevitably occur. Van de Moere and Ben Clark[17] reported a case of CSHRPE with a full-thickness macular hole in a 10-year-old boy. He hypothesized that fluid from the lesion could leak into the vitreous body. This further led to vitreous changes with premature liquefaction and degenerative changes. Tangential traction along the posterior vitreous was induced, subsequently leading to the development of a full-thickness macular hole.

However, whether the absence of a fascicular retinal nerve fibre layer is due to compression by the tumor is uncertain. Kita *et al*[2] reported a case of optic disc melanocytoma with concomitant fascicular retinal nerve fibre layer loss. The tumor was located on and above the surface of the optic disc. However, both the supratemporal and infratemporal retina demonstrated an absence of fascicular nerve fibre bundles. A dense vascular system was shown by OCTA in both the superficial and deep layers of the melanocytoma, which was clearly separated from the capillaries of the peripapillary retina. OCTA also showed reduced peripapillary perfusion in the area of the glaucomatous nerve fibre bundle defect. This finding suggested that structural damage is associated with glaucoma but not with optic disc melanocytic tumor compression. Most of the disease occurs in the fovea and adjacent to the foveola. However, in this patient, the lesion was located near the upper vascular arch and was accompanied by a loss of the bundled nerve fibre layer. Is the absence of fascicular nerve fibre bundles caused by the development of CSHRPE, or is it a concomitant combination of normal intraocular pressure glaucoma. There is a lack of relevant evidence to support this phenomenon, as no visual field examination was performed, and a longer period of observation is still needed to determine whether this increase has occurred.

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