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Optical coherence tomography of torpedo maculopathy in a patient with tuberous sclerosis

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A 12-year-old boy was diagnosed with tuberous sclerosis (TSC) and had a new deletion (c.1220_1240del21) in exon 11 in the TSC2 gene (Rendtorff et al. 2005). He is, to the best of our knowledge, the first patient diagnosed with this mutation. In addition to having tuberous sclerosis, the boy was born with bilateral upper limb reduction defect or meromelia. He was treated for epileptic seizures in his early childhood. His best-corrected visual acuity was 1.0/1.0. Ophthalmological examination revealed a single fundus lesion in the right eye and three in the left eye. Of the latter, two were ophthalmoscopically translucent and well-circumscribed, located at the inferior rim of the optic disc and near the temporal superior vascular arcade, respectively. The lesion in the right eye had comparable characteristics. The third lesion in the left eye was located temporal of the macula and had a horizontally elongated shape and a pale, well-defined appearance (Fig. 1A,B). On spectral domain optical coherence tomography (SD-OCT), the two aforementioned lesions in the left eye and the one in the right eye were confined to the retinal nerve fibre layer and transparent, while casting a weak shadow on the underlying retinal pigment epithelium and displacing the middle layers of the retina. One of them had a small intratumoural cavity (Fig. 2, panel A and panel B). The lesion (Fig. 2, panel C) in the left eye temporal of macula was confined to the subretinal space temporal of the fovea. It consisted of a large hyporeflective cavity between a thin retinal pigment epithelium (RPE) layer and a grossly attenuated or hypoplastic overlying photoreceptor inner and outer segment layer. There was increased signal transmission from the choroid corresponding to the lesion. The fundus lesions have remained unchanged during five years of follow-up.

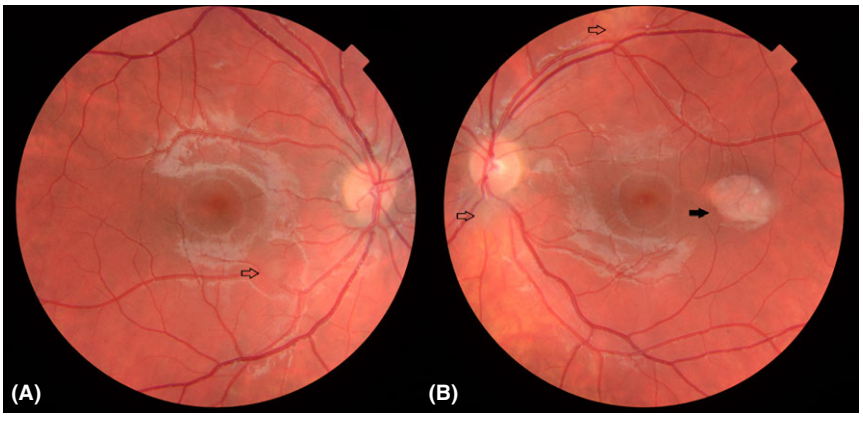

Fig. 1. A 12-year-old boy with tuberous sclerosis complex presented with bilateral astrocytic hamartomas and torpedo maculopathy in the left eye. (A) Fundus photo (right eye) showing an astrocytic hamartoma located near the inferior temporal vascular arcade. (B) Fundus photograph (left eye) showing two translucent and well-circumscribed astrocytic hamartomas, located at the inferior rim of the optic disc and near the temporal superior vascular arcade (arrows). The third lesion in the left eye, torpedo maculopathy, was located temporal of the macula and had a horizontally elongated shape and a pale, well-defined appearance (black arrow).

Phakomatoses, in general, are multisystem disorders with characteristic central nervous system, ocular and cutaneous lesions. Tuberous sclerosis is a rare autosomal dominant disease that causes hamartomas in different organs, including the brain, skin, kidneys, heart, lungs and the retina. Retinal astrocytic hamartoma is described as occurring in from 8% to more than half of the patients (Xu et al. 2012). Most astrocytic hamartomas are endophytic, meaning that they are confined to the retinal nerve fibre layer, but exophytic subretinal tumours can occur. Subretinal astrocytic hamartomas with intratumoural cavities have been visualized by SD-OCT (Shields et al. 2006). Hamartomas may resemble torpedo maculopathy, a rare, congenital, unilateral, hypopigmented lesion of the retinal RPE in the temporal macula (Wong et al. 2014). Torpedo maculopathy is an asymptomatic ovoid, and torpedo-shaped lesion with well-defined margins, and it is generally longer in the horizontal than in the vertical axis. Histopathological studies of torpedo maculopathy have, to our

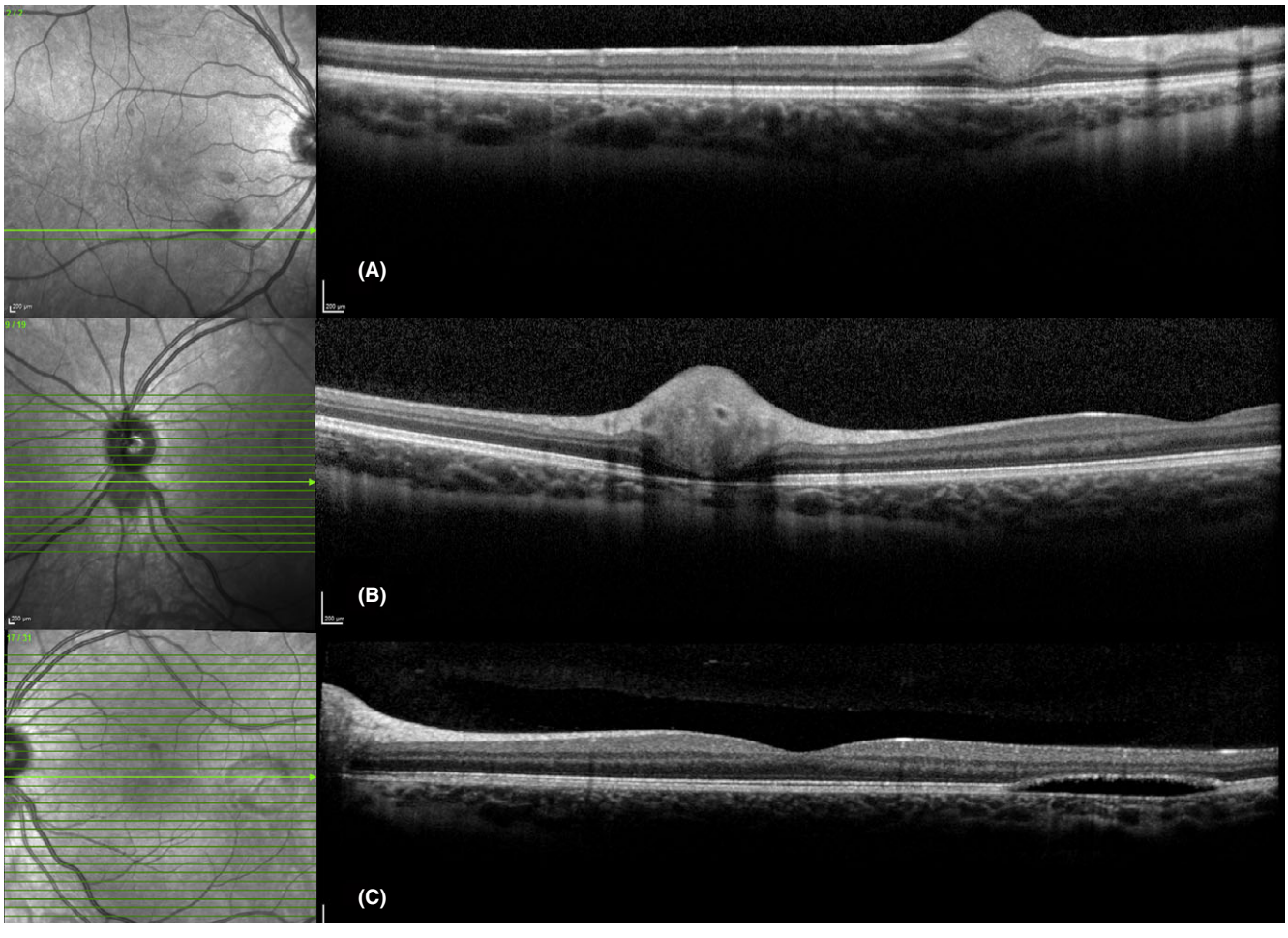

Fig. 2. Panel A (right eye): Spectral domain optical coherence tomography (SD-OCT) shows a transparent astrocytic hamartoma in the retinal nervefibre layer, displacing medial retinal layers, and located near the superior temporal vascular arcade. Panel B (left eye): SD-OCT shows an astrocytic hamartoma with a small intratumoural cavitation and displacing medial as well as outer retinal layers, and located at the rim of the optic disc. Panel C (left eye): SD-OCT shows torpedo maculopathy, located temporal of the fovea, with a large hyporeflective cavity between a thin retinal pigment epithelium (RPE) layer and a disrupted photoreceptor layer.

knowledge, never been reported. The aetiology remains unknown (Villegas et al. 2014). Other differential diagnoses include congenital hypertrophy of the RPE (CHRPE) and RPE lesions of Gardner syndrome. Here, we present a child with a torpedo maculopathy in tuberous sclerosis, a combination that has never before, to the best of our knowledge, been described. In our case, we have a patient with clinical and genetical verified TSC2, and classical endophytic astrocytic hamartomas who also presented with torpedo maculopathy in the left eye and bilateral upper limb meromelia, the latter of which has no known relation to tuberous sclerosis and which does not appear to be part of a known syndrome. We present the case to alert the scientific community to the existence of a case with such an unusual combination of clinical features that it prompts the speculation that it may have arisen not out of chance, but as a result of a hitherto unknown common aetiology that might be elucidated by the reporting of similar cases.

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