Torpedo Maculopathy Associated with Refractile Drusen and Age Related Macular Degeneration: Preserved Retinal Pigment Epithelial Function Around the Torpedo-Like Lesion?

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Abstract

Torpedo maculopathy (TM), a rare oval-shaped lesion in the temporal macula with its nasal tip toward the fovea, typically shows central hypoautofluorEscEncE with a hypHrautofluorHscHnt border suggesting retinal pigment epithelium (RPE) loss and dysfunction, respectively. Some of the optical coherence tomography (OCT) features described include neuro-retina thinning, outer retina disruption, RPE atrophy and a subretinal cOHi in some cases [1]. Recent multimodal imaging has revealed that intact RPE and Bruch's membrane on swept source OCT in some areas of the lesion [2]. We report an 84 y old Caucasian female with unilateral TM associated with intermediate dry age-related macular degeneration (AMD). To the best of our knowledge, the eldest TM case in the literature and the first with AMD. She was complaining of gradual deterioration in her reading vision and paracentral scotoma in the right eye. Uncorrected visual acuity was 6/9 right, 6/7.5 OHi Anterior segments were normal apart from lens nucleosclerosis. Funduscopy showed refractile drusen (material representing a byproduct of defective RPE-photoreceptor metabolism) with patchy areas of chorioretinal atrophy right eye). HH refractile drusen in the right macula were distributed within concentric zones across the macula, as recently described by Suzuki et al. [3]. In the OHi eye, the area around the torpedo lesion did not have refractile drusen or autofluorHscHncH features suggestive of RPE dysfunction. What is more, the OCT scans above and below the torpedo lesion revealed intact structure of the RPE/Bruch's membrane complex and inner choroid (Figures 1and 2). HH aetiology, pathophysiology and natural history of TM remain unknown. Various developmental defects have been proposed at the level of the horizontal raphe nerve fibHr layer [4], the RPE within the temporal bulge [5], and the emissary canal of the long posterior ciliary artery and nerve [6]. Here, we present a patient in her 9th decade with intermediate dry AMD where the RPE in the area around the torpedo lesion does not seem to have age-related RPE dysfunction to the same degree as the rest of the macular RPE in that eye or the fellow eye. HH pathogenesis of TM may possibly result in choriocapillaris, Bruch's membrane and/or RPE changes around the torpedo lesion that make this area less vulnerable to age-related dysfunction. Longer follow up will shed some light on how the outer retina structure and function evolves around the torpedo lesion.

Figure 1: Right eye macula color fundus photograph (a) showing medium-size refractile drusen in concentric zones around the fovea, mixed with areas of atrophy. HH OHi macula color photograph (b) shows a sharply demarcated temporal oval lesion along the horizontal raphe with the tip pointing towards the fovea, with outer hypopigmentation and inner hyperpigmentation. Medium-sized refractile drusen can be found in the nasal macula and inferiorly but not in the area around the torpedo-like lesion. Right eye autofluorHscHncH image (c) shows a mixture of hypoautofluorHscHncH indicating missing or dead RPE cells, and hypHrautofluorHscHncH corresponding with lipofuscin accumulation. /Hi eye autoflorHscHncH image (d) shows h\poautofluorHscHncH in the inner area of the torpedo-lesion and a mixture of hyper and hypoautofluorHscHncH in the nasal macula corresponding with the area where refractile drusen have developed.

Background: Torpedo maculopathy is a rare, congenital maculopathy classically diagnosed funduscopically as a 'torpedo-shaped' lesion located temporal to the fovea. This case describes a torpedo maculopathy with non-classic optical coherence tomographic (OCT) findings and collaborative OCT angiographic (OCTA) findings.

Case report: A 60-year-old Caucasian woman presented with a history of longstanding distortion and paracentral scotoma of the right eye. She had a positive family history of age-related macular degeneration. Visual acuity was 6/6 in each eye. Dilated fundus examination revealed a torpedo-shaped lesion in the right eye with a hypo-pigmented head pointing toward the fovea and a hyper-pigmented tail end. OCT imaging of the macula of the right eye revealed a subretinal cleft space with underlying thinning of the retinal pigment epithelium, increased choroidal reflectivity, as well as retinal pigment epithelial and choroidal excavation. OCTA choriocapillaris segmentation showed a hypo-reflective area associated with the lesion, adjacent to hyper-reflectivity. The patient was diagnosed with torpedo maculopathy of the right eye.

We report an 84 y old Caucasian female with unilateral TM associated with intermediate dry age-related macular degeneration (AMD). To the best of our knowledge, the eldest TM case in the literature and the first with AMD. She was complaining of gradual deterioration in her reading vision and paracentral scotoma in the right eye. Uncorrected visual acuity was 6/9 right, 6/7.5 OHi Anterior segments were normal apart from lens nucleosclerosis.

Funduscopy showed refractile drusen (material representing a byproduct of defective RPE-photoreceptor metabolism) with patchy areas of chorioretinal atrophy right!OHi eye). HH refractile drusen in the right macula were distributed within concentric zones across the macula, as recently described by Suzuki et al. [3]. In the OHi eye, the area around the torpedo lesion did not have refractile drusen or autofluorHscHncH features suggestive of RPE dysfunction. What is more, the OCT scans above and below the torpedo lesion revealed intact structure of the RPE/Bruch's membrane complex and inner choroid (Figures 1and 2).

Conclusions: OCT and OCTA imaging have been instrumental in developing a deeper understanding of many maculopathies, allowing for accurate diagnosis of macular conditions. Although the aetiology remains unclear, these imaging devices may provide further insight into the lesion in torpedo maculopathy.

Keywords: OCTA; optical coherence tomography; torpedo maculopathy.

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