Multimodality imaging of torpedo maculopathy with swept-source, en-face optical coherence tomography and optical coherence tomography angiography.

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Abstract

Purpose: Multimodality image analysis of two cases of torpedo maculopathy

Methods: Imaging with fundus photography, autofluorescence (AF), swept-source optical coherence tomography (SS-OCT), en face optical coherence tomography (En-face OCT) and optical coherence tomography angiography (OCTA).

Results: The basal diameter of the torpedo lesions was 1x2mm. One case had a satellite lesion. AF indicated variable loss of signal. SS-OCT and En-faceOCT demonstrated fundus excavation, attenuation of nuclear layers and disruption of the outer plexiform layer, loss of photoreceptors and a subretinal cleft. In one case, Sattler`s layer appeared extended at the cleft. OCTA indicated loss of flow in deep retinal vessels and increased flow in choroidal vessels surrounding the cleft.

Conclusions: Multimodal imaging findings of torpedo maculopathy include disruption of the deep retinal capillary network, expansion of Sattler`s layer and increased signal around the subretinal cleft.

Keywords

Torpedo maculopathy, swept-source, en-face, optical coherence tomography, angiography

Summary statement

Novel OCT findings are presented and discussed in two cases of torpedo maculopathy with the use of new imaging modalities namely swept source OCT and optical coherence tomography angiography with en-face OCT. These include the disruption of the deep capillary network, the expansion of Sattler`s network and increased signal around the subretinal cleft of lesions. Alterations of the choroidal vessels could be the result of a remodelling process.

Introduction

Torpedo maculopathy, an oval lesion in the temporal macula, is a congenital abnormality of the retinal pigment epithelium (RPE).[1] Modern imaging provides new insights on retinal anatomy. En-face OCT produces anteroposterior anatomic views following the contour of individual retinal and choroidal layers. Swept-source OCT (SS-OCT) uses longer wavelengths penetrating tissue to a greater extent than conventional OCT allowing imaging of the choroid with superior resolution.[2] Optical coherence tomography angiography (OCTA) allows non-invasive visualization of retinal and choroidal vasculature, mapping erythrocyte movement over time by comparing sequential OCT B-scans.[3] We present the multimodal imaging analysis of two torpedo maculopathy cases.

Patients and methods

A 24-year-old Afrocarribean male and a 57-year-old Asian female patient were referred to the Medical Retina Service of Moorfields Eye Hospital, following the incidental finding of a pigmented lesion at the left macula. Best-corrected visual acuity was 6/6 in each eye. Prior ophthalmic, medical or family histories were negative. Fundus examination revealed the presence of an oval, partially pigmented lesions consistent with torpedo maculopathy. Multimodality imaging was undertaken with fundus photography, AF scanning (Spectralis, Heidelberg Engineering, Dossenheim, Germany), SS-OCT (DRI-OCT Atlantis,Topcon Medical Systems, Oakland, New Jersey, USA), En-face OCT and OCTA (AngioVue, Fremont, CA, USA).

Results

Fundus images (Fig.1a/2a) demonstrated the oval lesion temporal to the fovea(1x2 mm), the nasal aspect of which was de-pigmented and the lateral third heavily pigmented. On red-free image (Fig.1b) a much smaller (0.3x0.3 mm) satellite lesion was discernible in one case. AF (Fig.1c/2b) showed variable loss of autofluorescence throughout both lesions.

SS-OCT (Fig.1d/2c sections 1-3) over the lesion demonstrated the presence of fundus excavation. At this point, attenuation of the nuclear layers with disorganisation of the outer plexiform layer and the myoid, ellipsoid and interdigitation zones were noted. The RPE-Bruch`s membrane complex appeared thickened, hyperreflective with a subretinal cleft. The cleft was more accentuated in the first case in which Sattler`s layer appeared to project deeper and laterally towards Haller`s layer; this correlated topographically with loss of anatomic integrity of the ellipsoid zone. The satellite lesion appeared to have similar features (Fig.1d-3). With En-face OCT (Figs.1,2) delineation of defects at the RPE level and attenuation changes at the level of the internal limiting membrane were noted.

Juxtaposition of En Face OCT with OCTA (Figs.1,2) revealed additional findings, namely attenuation of flow of the deep retinal vascular network at the level of the cleft with hyperreflectivity of vessels around it.

Discussion

In this report we present the multimodality imaging features of torpedo maculopathy, for the first time using SS-OCT, en-face OCT and optical coherence tomography angiography (OCTA). Torpedo maculopathy, has been described as a congenital malformation of the retinal pigment epithelium, of variable size and pigmentation.[4-8] It was also termed paramacular coloboma, but these typically feature loss of retinal layers, a feature not demonstrated in torpedo maculopathy.[4]

The etiology of torpedo maculopathy remains speculative: theories include incomplete differentiation of the arcuate nerve fiber bundles along the horizontal raphe,[4] defect in the development of the RPE within the fetal temporal bulge [6] or a malformation of the emissary canal of the long posterior ciliary artery and nerve.[7] Rare associations with blepharophimosis or retinal vessel situs inversus exist, though our cases had no associated findings.[4] **Despite the novel findings**

provided by new imaging technologies no frank association can be attempted with the existing pathogenetic theories.

Satellite lesions in the direct vicinity of the main lesion can be found, as well as areas of atrophy contiguous or adjacent to the tail portion.[7,9-10] The presence of a distinct satellite lesion may indicate disturbance of chorioretinal differentiation, but could also represent a forme-fruste manifestation of variable penetrance.[10]

Imaging

There has been a recent attempt to classify spectral domain OCT findings in torpedo maculopathy, pertaining to the outer retinal layers. [8] Cases were divided into Type I with thinning of the outer nuclear layer, **attenuation** of interdigitation and the ellipsoid zone but without outer retinal cavitation and Type II cases with both attenuation and cavitation of outer retinal structures. The current cases fall into Type II by this classification. Cystic degeneration of the inner retinal layers is a commonly reported feature,[5] not encountered in the present cases.

OCTA indicated the attenuation of retinal vasculature in the deep retinal vascular network. This loss was associated with the outer retinal layer and RPE changes. This is a novel finding as imaging of retinal vascular networks was previously unavailable and beyond the scope of fluorescein angiography.[4,10]. **Unfortunately, quantitative means for the vascular density are not available**.

SS-OCT demonstrated choroidal excavation, hypertrophied RPE cells and intact Bruch's membrane, in keeping with other reports.[4,5] OCTA demonstrated increased density of choroidal vessels around the cleft. OCTA signal strength is related to erythrocyte movement, thus this is interpreted as increased flow. However, **this may also be due to mismatch between blood cell velocity and OCT instrument speed [3]. Most of the OCTA signal in the choroid is generated from choroidal stroma and in these cases, they may be**

enhanced through a transmission defect in and around the torpedo lesion.

On SS-OCT this vascular rearrangement was demonstrated by expansion of the Sattler`s layer in one case. This is an interesting finding, with further studies required to confirm this. Histologic studies are unavailable on torpedo maculopathy. Spectral domain OCT showed atrophic choriocapillaris in a type I lesion, which may also indicate choroidal change at the point of retinal attenuation.[9] .

AF signal loss is either homogeneous or heterogeneous, focused on the nasal or temporal portion of the lesion, and not related to the degree of pigmentation.[8] In the current cases, there is mild hyperautofluorescence towards the nasal portion of the lesion. This area showed gradual attenuation of the ellipsoid and interdigitation zones on SS-OCT, indicating a declining interaction of the RPE with the photoreceptor layer. That interaction was lost further temporally in the lesion, and the AF signal completely lost.

In conclusion, findings of loss of deep retinal vessels and **potential remodelling of** choroidal vessels in conjunction with fundus excavation and disorganisation of the neurosensory retina were found in torpedo maculopathy with SS-OCT and OCTA. It is unclear whether the choroidal vessel alterations are a late-onset phenomenon to the retinal malformation or whether they constitute a primary event.

1031 words

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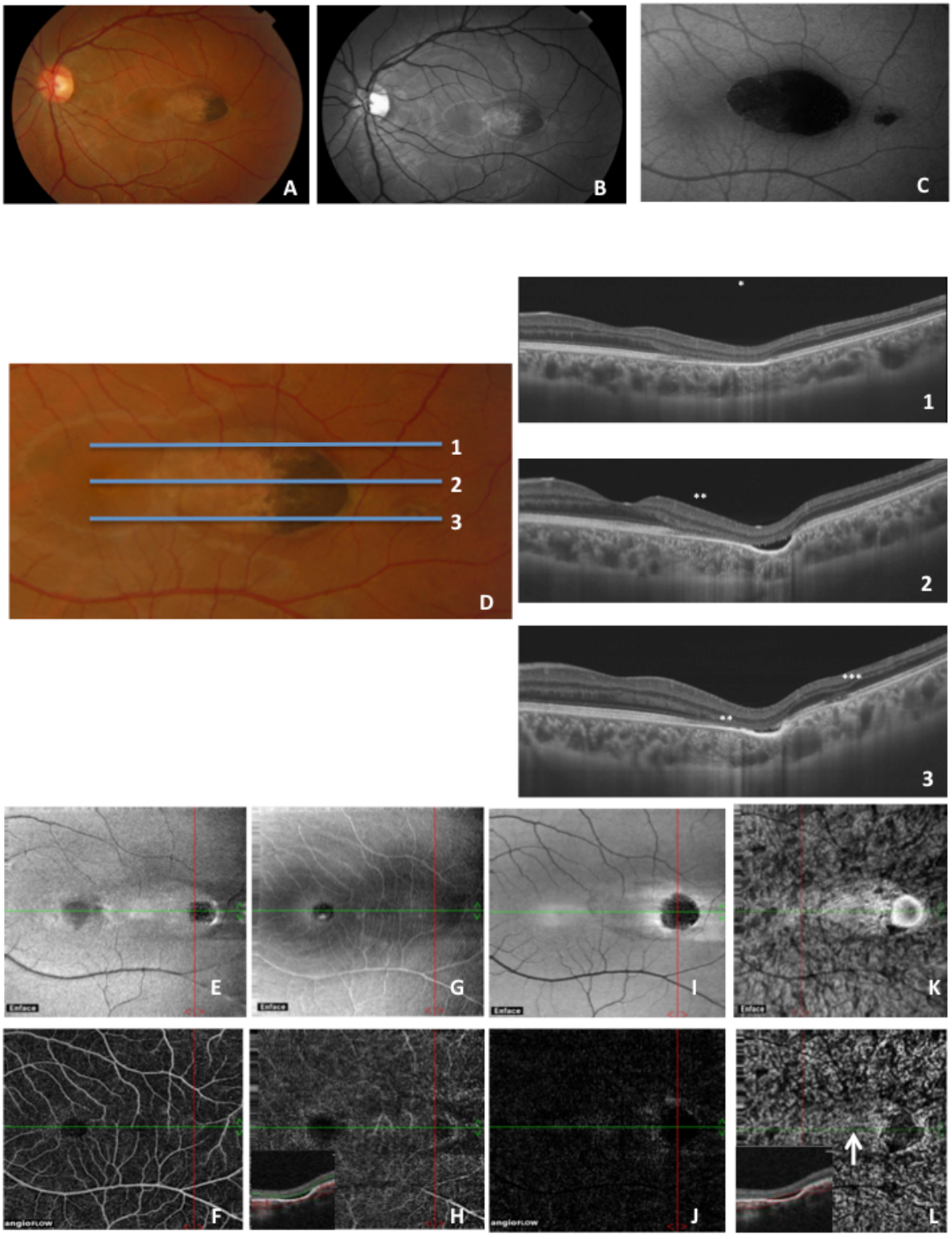
FIGURE LEGENDS

Fig 1. Multimodal imaging analysis of the left macula of a 24-year old patient with torpedo maculopathy. (a,b) **Fundus photograph and red free image** demonstrating the presence of an oval depigmented lesion located temporally to the fovea (c) **Autofluorescence scan** demonstrating loss of autofluorescence and a satellite lesion adjacent to the main lesion. (d) **Swept-source optical coherence tomography (SS-OCT)** demonstrated the presence of fundus excavation. (d1) Superior edge of the lesion. Thinning of ganglion cell layer (*) and reduction of the inner and outer nuclear layers with evident disorganisation of the outer plexiform layer and the myoid, ellispoid and interdigitation zones. The RPE-Bruch complex intact with increased signal. Choriocapillaris` and Sattler`s layer signals appear enhanced with loss of Haller`s layer up to the level of choroid-sclera interface. (d2) Centermost area bears complete loss of the outer nuclear layer. At the level of excavation, myoid. ellipsoid and intedigitation zones appear compacted. The RPE-Bruch complex appears thickened, hyper reflective and forms a subretinal cleft. Sattler`s layer appears to project deeper and laterally towards Haller`s layer (**); this expansion correlating superiorly to the point where there is loss of the anatomical integrity of the zones.(d3) Satellite lesion (***) with minimal fundus excavation. The inner nuclear and the outer plexiform layers follow the excavation pattern with reduced thickness of the outer nuclear layer. The external limiting membrane is intact though displaced, the myoid, ellipsoid and interdigitation notes are again disrupted. At the level of the choroid, the signal appears increased at the level of choriocapillaris and Sattler`s layer. **Juxtaposition of En Face OCT with optical coherence tomography angiography (OCTA)** (e-l). Imaging at the level of the superficial retinal vascular network (e,f) did not reveal vascular changes despite the fact that on en face OCT the fundus excavation is discernible. At the deeper retinal capillary vascular network (g,h) loss of vessels at the point of the cleft is demonstrated (segmentation line sinset at h). The outer retinal layer demonstrates(i,j) foci of increased vessel reflectivity all corresponding to areas within the lesion and surrounding the cleft. At the level of choriocapillaris (k,l) increased signal of choroidal vessels surrounding the cleft (arrowhead). (inset in l segmentation lines)

Fig 2. Multimodal imaging analysis of the left macula of a 57-year old patient with torpedo maculopathy. (a) **Fundus photograph** demonstrating the presence of an oval depigmented lesion located temporally to the fovea (b) **Autofluorescence scan** demonstrating loss of autofluorescence primarily at the temporal aspect. (c) **Swept-source optical coherence tomography (SS-OCT)** at the site of the lesion demonstrated the presence of fundus excavation. (c1-3) Reduction of the inner and outer nuclear layers with evident disorganisation of the outer plexiform layer are noted. The external limiting membrane appears intact. At the point of neuroretinal excavation, there is disorganization of the myoid, ellispoid and interdigitation zones with the formation of a subretinal cleft. The RPE-Bruch complex appears intact but its signal increased. The choriocapillaris and Sattler`s layer signals appear slightly enhanced **Juxtaposition of En Face OCT with optical coherence tomography angiography (OCTA)** (d-k). Superficial retinal vascular network (d,e) did not reveal vascular changes though fundus excavation is discernible on en-face OCT. At the deeper retinal capillary vascular network (f,g) attenuation of vessels at the point of the cleft is demonstrated (inset in g – segmentation lines). The outer retinal layer demonstrates(h,i) foci of increased vessel reflectivity all corresponding to areas within the lesion and surrounding the cleft. At the level of choriocapillaris (j,k) loss of choriocapillaris with increased reflectivity of choroidal vessels surrounding the cleft (arrowhead) (inset in k – segmentation lines)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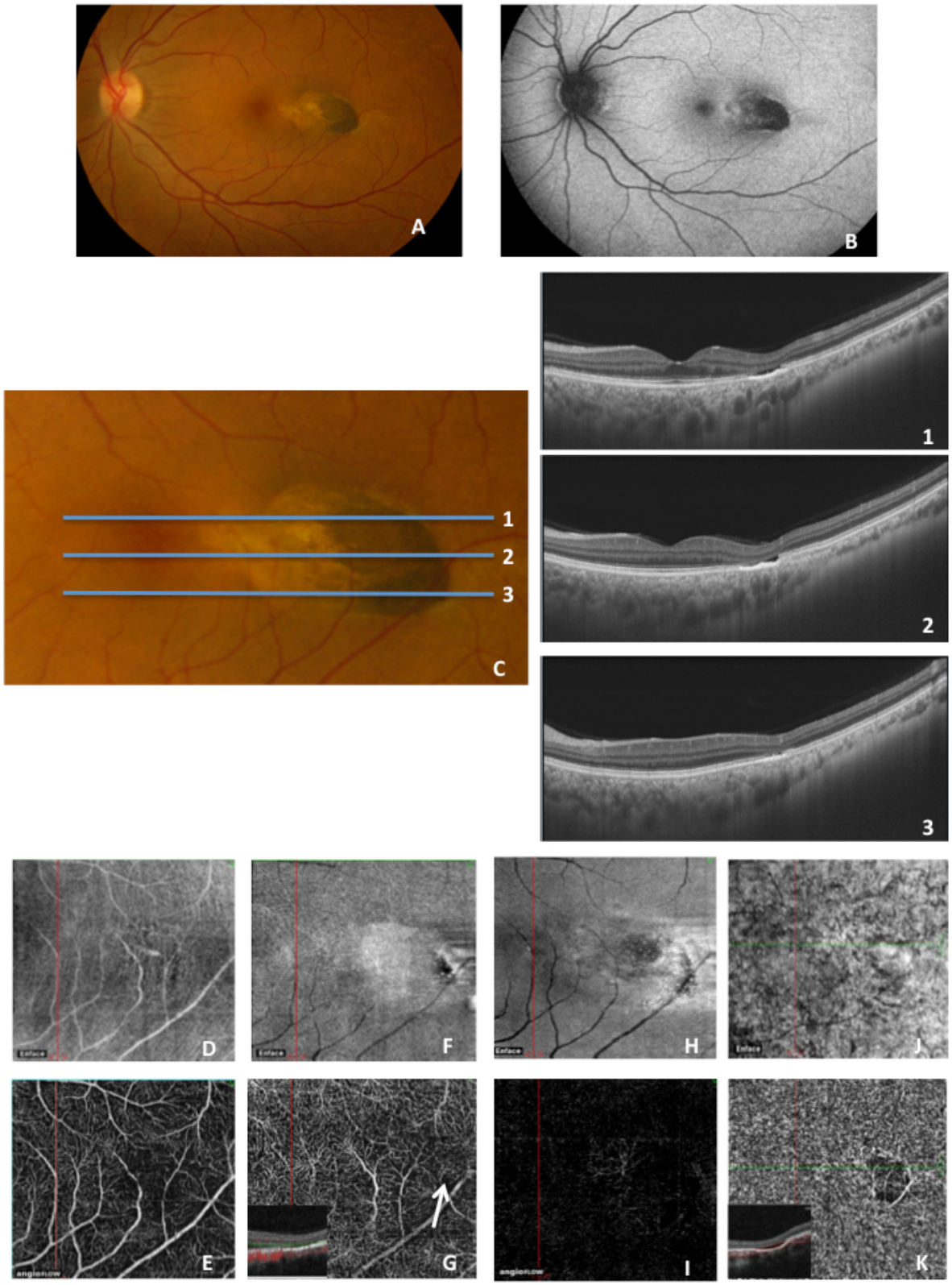
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