

A novel phenotype of torpedo maculopathy on spectral-domain optical coherence tomography

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ABSTRACT

Purpose: To present a new phenotype of torpedo maculopathy on spectral domain optical coherence tomography imaging (SD-OCT).

Observations: A 31-year-old female presented with a multi-partite yellowish lesion in the macula of her left eye, with a central fovea-involving component and a temporal tail-like component. The lesion showed mixed hyper- and hypoautofluorescence on fundus autofluorescence imaging. The fovea-involving component exhibited disruption of ellipsoid zone without outer-retinal cavitation on SD-OCT, consistent with the prior-described type 1 OCT morphology. The temporal tail showed subtle inner choroidal excavation with preservation of the ellipsoid zone and outer-retinal structures.

Conclusions: Inner choroidal excavation with preservation of the overlying outer-retinal structures represents a novel morphological phenotype on SD-OCT in torpedo maculopathy. This case demonstrates that distinct morphological subtypes may co-exist in different regions of the same torpedo maculopathy lesion.

1. Introduction

Torpedo maculopathy, originally described as a “Hypopigmented Nevus of the RPE” by Roseman and Gass¹ in 1992, is a unilateral congenital developmental abnormality of the retinal pigment epithelium and/or choroid with stereotypical morphology and topography. A variably pigmented lesion, classically located in the temporal macula with rare foveal involvement and exhibiting a characteristic teardrop or “torpedo-like” shape with apex oriented toward the fovea, it has been hypothesized to be the result of a number of possible aberrant processes, including a defect in RPE development at the temporal fetal bulge,² malformation of an emissary canal,³ or incomplete arcuate bundle differentiation.⁴ Wong et al.⁵ described two morphologic phenotypes into which torpedo maculopathy lesions can be divided based on optical coherence tomography (OCT) imaging: type 1, lesions demonstrating outer retinal attenuation without cavitation, and type 2, lesions exhibiting outer retinal attenuation and cavitation with or without inner choroidal excavation. Tripathy et al.⁶ recently expanded the classification system in 2018, adding a case in which choroidal excavation was accompanied by degeneration of the outer retina and a subretinal cleft; they proposed such a phenotype be labeled as type 3. Since Wong et al.’s report the majority of subsequent case reports have adopted this general

categorization system,^{7–9} but it is not clear whether this system is exhaustive in describing the spectrum of possible OCT morphologies inherent to this clinical entity. Herein, we present a case of torpedo maculopathy with unique OCT findings not consistent with the prior proposed type 1–3 phenotypes.

1.1. Case report

A 31-year-old female with history of depression, migraine, and non-ocular Lyme disease was referred to the retina service of the Johns Hopkins Wilmer Eye Institute for a “freckle” in the fundus of the left eye that was noted at least 10 years prior. The patient reported chronic distortion in the left eye nasal to fixation. The patient denied any family history of inherited retinal disease, other ocular illnesses, or history of ocular trauma.

Examination of the right eye showed a completely normal examination with a Snellen acuity of 20/20. Examination of the left eye was notable for a Snellen acuity of 20/40, normal anterior segment exam, lack of intraocular inflammation, normal peripheral retina, and a lesion in the macula. The yellowish lesion was centered on the fovea, with a tail-like structure extending to the temporal macula (Fig. 1) and abnormal hyper- and hypoautofluorescence signals in the same area on

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Fig. 1. Color fundus photograph of the patient's left eye: a yellowish macular lesion with a tail-like structure extending to the temporal macula, consistent with torpedo maculopathy. (For interpretation of the references to colour in this figure legend, the reader is referred to the Web version of this article.)

fundus autofluorescence imaging (Fig. 2).

Based on the clinical appearance, a diagnosis of torpedo maculopathy was made. In addition, the torpedo maculopathy lesion was imaged with spectral domain OCT, which revealed two patterns. Line scan through the fovea portion of the lesion showed loss of the ellipsoid zone but no outer retinal cavitation (Fig. 3A). Line scan through the temporal tail portion of the lesion showed subtle inner choroidal excavation, but the overlying ellipsoid zone remained intact (Fig. 3B).

2. Discussion

Herein, we present a case of torpedo maculopathy which, to our knowledge, contains novel OCT features not previously described in the literature. Of note, our patient specifically denied any history of ocular trauma, and similar multi-partite lesions with a temporal tail-like

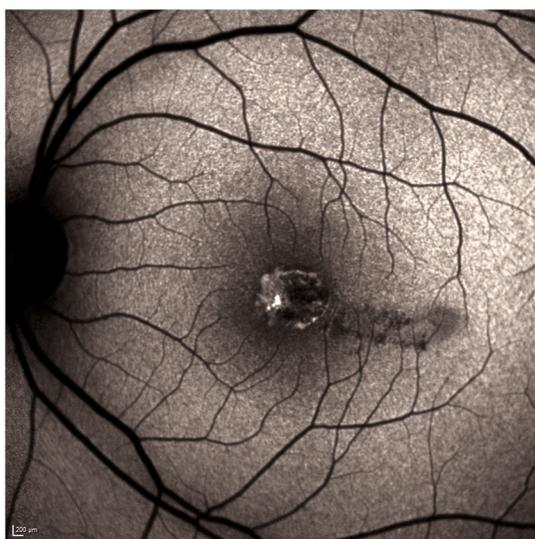


Fig. 2. Corresponding fundus autofluorescence imaging, revealing mixed areas of hyperautofluorescence and hypoautofluorescence.

structure have been previously reported in cases of torpedo maculopathy.^{5,8,10} The foveal involvement of our patient's central lesion is somewhat atypical for torpedo maculopathy. However, torpedo maculopathy with foveal involvement has been reported before by Angioi-Duprez and Maalouf in 2000.¹¹

Based on the OCT classification by Wong et al.,⁵ recently expanded by Tripathy et al.,⁶ torpedo maculopathy can be categorized into 3 types. Type 1 involves outer retinal attenuation without cavitation, while type 2 involves both outer retinal attenuation and outer retinal cavitation with or without inner choroidal excavation. Type 3 is essentially a severe type 2 lesion with additional features of inner-retinal surface cavitation and an acute subretinal cleft. Therefore, the fovea portion of our lesion can be classified as type 1. However, the temporal tail portion of our lesion shows preservation of the ellipsoid zone and lack of subretinal fluid despite the presence of inner choroidal excavation, thus not fitting the description of either type 1, type 2, or type 3 lesions. As such, we propose that this particular configuration represents a new type 4 morphology that has not yet been described before. In addition, our case suggests that multiple OCT morphologic subtypes can co-exist in different geographic areas of the same lesion, an observation that was not noted in the review and case series by Wong et al.⁵

There has been debate as to whether these different morphologies are representative of chronological evolution of torpedo lesions or are truly independent phenotypes. Wong et al. proposed the former, noting that most type 2 lesions were typically observed in older patients. However, Shirley et al.⁸ noted the definite presence of type 2 lesions in pediatric patients in their case series, casting some doubt about the chronological sequence hypothesis.

Recent publications have further characterized various torpedo lesions using OCT angiography (OCT-A).^{7,9,12–14} OCT-A findings included preservation of superficial capillary plexus perfusion and flow loss in the deep capillary plexus and in the area of subretinal clefting. Grimaldi et al.¹³ further found reduced sensitivity on microperimetry corresponding to the area of a torpedo lesion, confirming an abnormal functional component in this clinical entity. A weakness of our current report is a lack of OCT-A and microperimetry findings to further deepen our characterization of this novel type 4 morphology. Future case reports of type 4 lesions that present such findings would be welcome and most informative.

3. Conclusions

In summary, our case of torpedo maculopathy extends the OCT morphological spectrum of this clinical entity. Specifically, preservation of the ellipsoid zone, lack of subretinal fluid and choroidal excavation can co-exist, a morphologic phenotype that we propose as "type 4." It is difficult to make sweeping conclusions based on one case, and it remains unknown how often such "type 4" configuration is seen in torpedo maculopathy. Also, future studies with longitudinal registered OCT line scans can help elucidate whether type 1–4 morphologies remain static or whether they represent different stages of evolution over time.

Patient consent

Consent to publish the case report was not obtained. This report does not contain any personal information that could lead to the identification of the patient.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

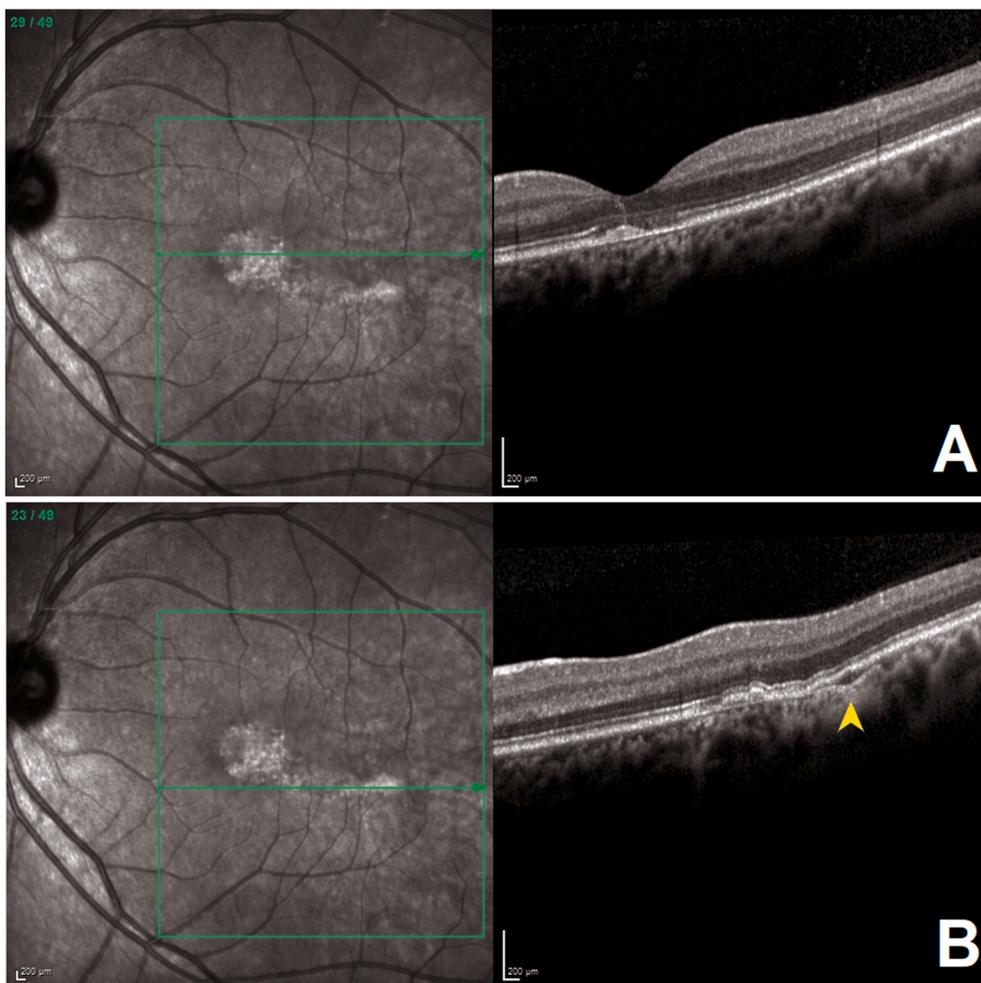


Fig. 3. SD-OCT. Foveal portion of the lesion showing disruption of ellipsoid zone without outer retinal or inner choroidal cavitation (A). Temporal tail of the lesion showing intact ellipsoid zone and localized inner choroidal excavation (arrowhead) without subretinal fluid (B).

Other contributions to present work: None.

Declaration of competing interest

None of the authors have any financial disclosures.

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