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A unique presentation of subretinal fluid in a type III torpedo maculopathy phenotype

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ABSTRACT

Purpose: To report a rare type III torpedo maculopathy lesion with a unique manifestation of subretinal fluid.
Observations: A nine-year-old patient was referred to retina for an evaluation of a hypopigmented oval-shaped lesion in the temporal macula with an area of inferior subretinal fluid in the right eye. The lesion demonstrated inner and outer retinal and retinal pigment epithelial attenuation, intraretinal and subretinal fluid, a serous neurosensory retinal detachment, and inner choroidal excavation on optical coherence tomography. Fundus autofluorescence showed a lane of downward-tracking fluid. Intravenously administered fluorescein angiography revealed a window defect in the area of the torpedo lesion suggesting choroidal flush.
Conclusions and Importance: The case is the third documented case of torpedo maculopathy with subretinal fluid in the literature with a unique combination of intraretinal cystic changes and dependent descending subretinal fluid, somewhat akin to a Best disease outside of the fovea with choroidal excavation. The morphology of torpedo maculopathy continues to expand as more cases are revealed.

1. Introduction

Torpedo maculopathy is a rare congenital condition of the retinal pigment epithelium (RPE) first described by Gass and Roseman in 1992.[1] It is described as a horizontal torpedo-shaped hypopigmented lesion found in the temporal macula with a nasal wedged tip pointed towards the optic disc and a frayed or rounded temporal tail.[2] Although largely asymptomatic, torpedo maculopathy has been linked in rare instances with mild to moderate vision loss, scotomas, and choroidal neovascularization.[3,4]

With fewer than 100 reported cases in the literature, the full scope of presentation of torpedo maculopathy is yet to be determined. The advent of multimodal imaging has allowed for the evolving characterization of its features as more is learned of the disease. In this report, we describe a unique case of torpedo maculopathy characterized by a concomitant presentation of subretinal fluid with unique features yet to be described in the literature.

2. Method of literature search

The NIH Pubmed Database was searched for "torpedo maculopathy," returning 87 published papers that were reviewed for context and comparison. While there were no cases precisely matching the presentation of the enclosed patient, the papers detailing subclassification of torpedo maculopathy with subretinal fluid proved useful.

3. Case report

A healthy nine-year-old female with a history of attention deficit hyperactivity disorder was referred to the retina clinic for evaluation of possible ocular melanoma in the right eye. She reported seeing colorful red and green lights in the evening. A "black spot" in her right eye was noted at her last optometry visit. The patient's own and familial ocular history were otherwise unremarkable.

Best corrected visual acuity was 20/25 in the right eye and 20/20 in the left eye. Intraocular pressures, pupils, confrontational visual fields, and extraocular motility were normal. Bilateral anterior segment exams and dilated fundus exam of the left eye were normal. Retinal examination of the right eye showed an oval-shaped hypopigmented lesion three disc diameters in size and immediately temporal to the fovea with an area of adjacent inferior subretinal fluid (Fig. 1). A corresponding area of hypoautofluorescence with a surrounding margin of hyperautofluorescence most concentrated at the inferior border was found on fundus autofluorescence, suggestive of dependent collection of shed

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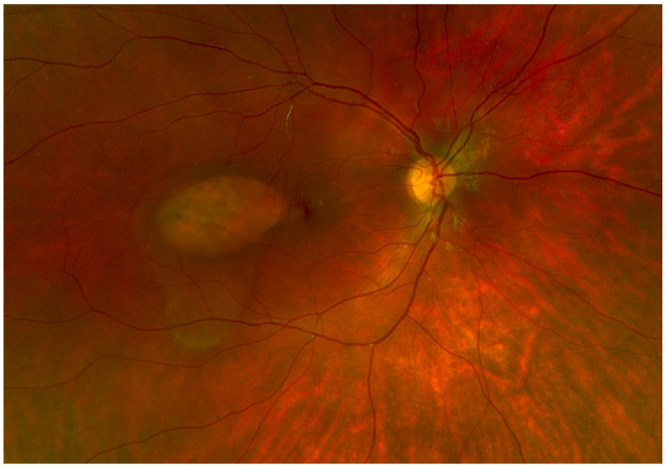

Fig. 1. Widefield color fundus photography of the right eye illustrated a horizontal hypopigmented oval lesion in the temporal macula with inferior subretinal fluid.

outer segments (Fig. 2). Optical coherence tomography (OCT) illustrated attenuation of the interdigitation and ellipsoid zones, outer nuclear layer, and retinal pigment epithelium; inner choroidal excavation; intraretinal and subretinal fluid; and a neurosensory retinal detachment causing focal elevation at the site of the lesion (Fig. 3). Interestingly, the outer segments elongated to a stalactite configuration reminiscent of Best disease.[5] Critically, fluorescein angiography (FA), administered intravenously rather than orally, demonstrated window defect without leakage with hyperfluorescence (Fig. 4).

4. Discussion

Multiple theories have been proposed regarding the pathogenesis of torpedo maculopathy. Sharma et al. suggested the lesions arise from faulty differentiation or maldevelopment of the retina based on histologic evidence of the absence of RPE and choriocapillaris.[6] Tietelbaum et al. and later Papastefanous et al. proposed the prenatal disturbance of choroidal or short posterior ciliary vasculature disrupting the RPE.[7,8] Pian et al. proposed a defect of the architecture along the arcuate nerve fiber layer bundles during development of the horizontal raphe.[9] Shields

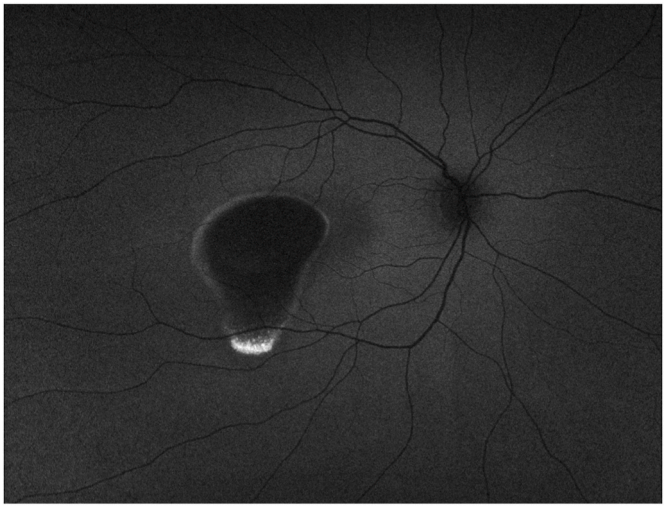

Fig. 2. Fundus autofluorescence showed a corresponding hypoautofluorescent oval lesion and descending tract of subretinal fluid. The surrounding margin is hyperautofluorescent, suggestive of lipofuscin produced by dysfunctional retinal pigment epithelial cells or those under metabolic stress.

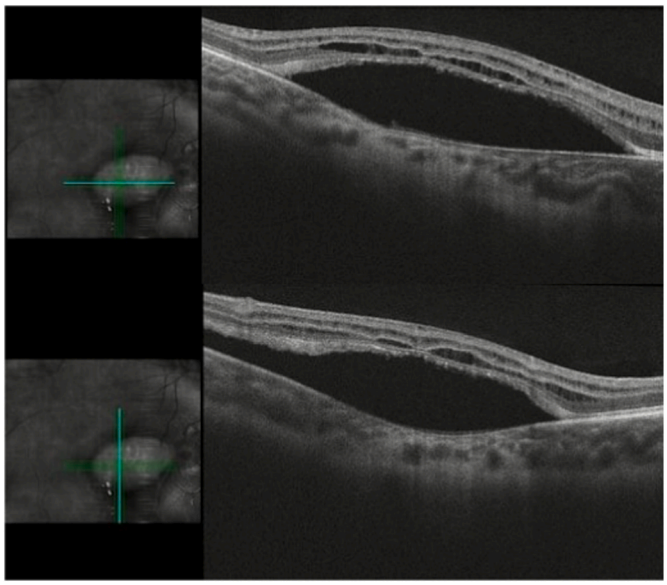

Fig. 3. Horizontal and vertical optical coherence tomography demonstrated inner retinal cystic changes, outer segments elongated to a stalactite configuration, retinal pigment epithelial attenuation, subretinal fluid noticeably tracking inferiorly, and inner choroidal excavation.

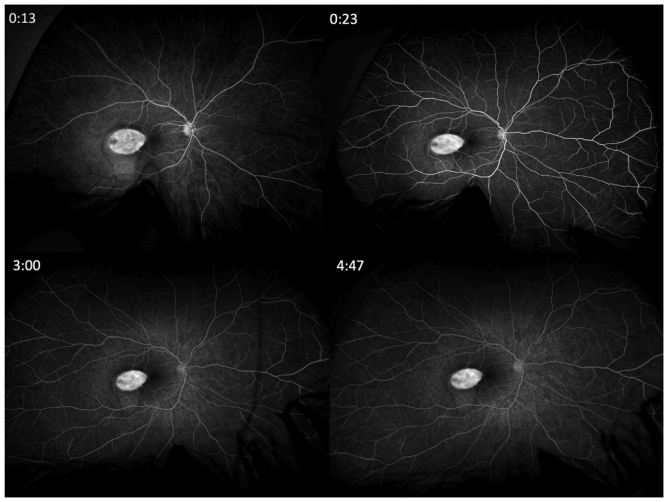

Fig. 4. Fluorescein angiography showed window defect as early hyperfluorescence corresponding to choroidal flush remained at the same intensity as the imaging progressed.

et al. proposed torpedo lesions are defects of the fetal temporal bulge, a "cone-shaped bulge" within the posterior pole centered just temporal to the fovea that expands the macular area by 8-months gestation according to fetal RPE development studies.[10] da Manuel-Triantafilo et al. related torpedo maculopathy to sequelae of intrauterine chorioretinitis.[11] Most recently, Hugo et al. demonstrated alteration of the cone mosaic on multimodal imaging with adaptive optics that could be explained by RPE alteration, lending support to a persistent defect in the RPE as the etiology.[12]

Although exact etiology is still unknown, OCT has allowed for characterization of the spectrum of findings of torpedo maculopathy and its classification. Wong et al. first identified two abnormal lesion patterns, type I with outer retinal attenuation and type II with both outer retinal attenuation and cavitation with or without inner choroidal excavation.[13] Tripathy et al. later described lesions with outer retinal cavitation with subretinal clefts as type II variants and classified type II

excavated lesions with additional features – retinal thinning and inner hyperreflective layers but without subretinal clefts – as type III.[14] Most recently, a case with preserved retinal structure in the presence of solely choroidal excavation was deemed type IV.[15]

Herein, we report the third known case of torpedo maculopathy associated with sub-retinal fluid. Panigrahi et al. and Kerwat et al. have reported the others.[16,17] Both aforementioned cases show neurosensory retinal detachments larger than the subretinal clefts found in typical lesions, and the Panigrahi et al. case contains intraretinal cystic changes absent from typical lesions. However, our case uniquely demonstrates a gravitational tract of fluid readily seen on multimodal imaging, a feature of chronicity found in a minority of central serous chorioretinopathy cases.[18,19] Our lesion on OCT possesses outer retinal attenuation alongside outer segment elongation and shedding, outer choroidal excavation, prominent retinal detachment with expansive subretinal fluid, and rare intraretinal retinoschisis. With a combination of type I and II characteristics, we present a type III torpedo maculopathy with a unique presentation of subretinal fluid.

Regarding management, the patient was offered topical dorzolamide to treat cystoid macular edema; however, the patient and family elected for observation given excellent visual acuity.

5. Conclusions

In summary, our case report adds to the growing spectrum of torpedo maculopathy phenotypes and provides another variant of a type III lesion, specifically one with subretinal fluid. Where this expression falls along the spectrum is unknown given the scarcity of knowledge of this disease. Determining if these cases represent progressive changes of one another or separate entities will require longitudinal follow-up studies.

Patient consent

Consent to publish case report was not obtained. This retrospective report does not contain any protected health 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.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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