DOI: 10.14744/eer.2021.76486
Eur Eye Res 2021;1(1):53–56

EUROPEAN EYE RESEARCH

CASE REPORT

Torpedo maculopathy: A single entity with three different presentations

Ceren Durmaz Engin,[1] Ziya Ayhan,[2] Ayse Tulin Berk,[2] Ali Osman Saatci[2]

[1]Department of Ophthalmology, Karadeniz Eregli State Hospital, Zonguldak, Turkey
[2]Department of Ophthalmology, Dokuz Eylul University Faculty of Medicine, Izmir, Turkey

Abstract

Torpedo maculopathy (TM) is a benign and non-progressive congenital lesion of the retina pigment epithelium in association with the disruption of outer retinal layers. In this case series, three patients with unilateral torpedo lesions who displayed different clinical features were reported. In all cases, there was somewhat distortion of the outer retinal layers with a corresponding increase in the choroidal reflectance under the lesion in optical coherence tomography (OCT). Fluorescein angiography and OCT angiography were performed in the only adult case. As TM is mostly a benign entity without causing any visual disturbance, its differential diagnosis carries paramount importance.

Keywords: Hypopigmented lesion; multimodal imaging; optical coherence tomography; retina pigment epithelium; torpedo maculopathy.

Torpedo maculopathy (TM) is a rare, solitary hypopigmented lesion of the retina pigment epithelium (RPE) located in the vicinity of the macula mostly without causing any significant visual deficit. The condition was described as albinotic nevi of RPE, congenital hypomelanotic freckle, and solitary amelanotic spot by several authors but a unilateral "torpedo-shaped" hypopigmented lesion, typically located temporal to the macula with a nonprogressive course is the common presentation.[1,2] Although it is well-recognized by its characteristic shape, the underlying pathophysiological process is not clearly known yet. Several theories have been proposed as of the etiology, including a dysgenetic RPE, a developmental defect in the "fetal temporal bulge", abnormalities of underlying choroid vasculature, or failure of the RPE to close the overlying the region near the emissary canal of ciliary vessels.[3,4] Visual acuity (VA) is typically not affected. Although progression is not expected, these lesions are fraught with macular neovascularization (MNV) occurrence due to RPE and outer retinal damage and should be regularly monitored.[5] In our case series, we report three patients with TM presented with different clinical features.

Case Report

Case 1– A 2-year-old Caucasian girl was referred to our clinic for suspicion of squint presence. Her VA was recorded

Cite this article as: Durmaz Engin C, Ayhan Z, Berk AT, Saatci AO. Torpedo maculopathy: A single entity with three different presentations. Eur Eye Res 2021;1:53-56.

Correspondence: Ali Osman Saatci, M.D. Department of Ophthalmology, Dokuz Eylul University Faculty of Medicine, Izmir, Turkey
Phone: +90 232 390 49 08 **E-mail:** osman.saatci@gmail.com
Submitted Date: 18.03.2021 **Accepted Date:** 05.04.2021

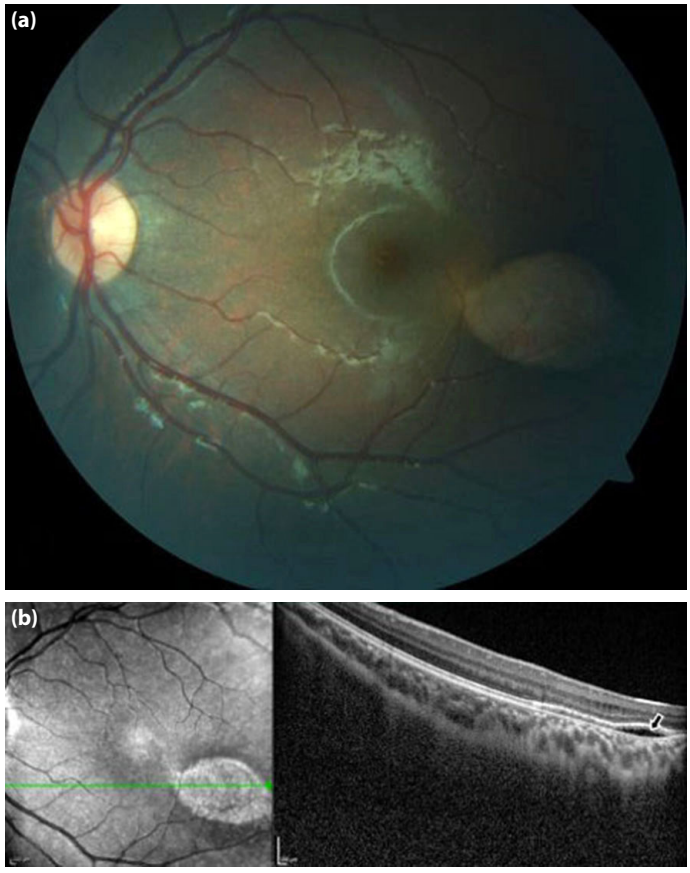

Fig. 1. **(a)** Color fundus photo of left eye demonstrates a flat, torpedo-shaped hypopigmented lesion with a tail pointed toward the fovea. **(b)** Enhanced depth imaging optical coherence tomography scan shows outer retinal layer disorganization accompanied by a subretinal cleft (arrow) and increased reflectivity of the choroid underneath.

as fix and follow and central, steady, maintained bilaterally. Her anterior segment evaluation and ocular alignment were unremarkable, and her cycloplegic refraction was within normal limits for her age group. Fundus examination revealed a torpedo-shaped hypopigmented lesion with a well-defined margin temporal to the left macula (Fig. 1a). Enhanced depth imaging optical coherence tomography (EDI-OCT) scan through the lesion showed distortion of the outer retina with increased reflectivity of the choroid together with a subretinal cleft (Fig. 1b). She was diagnosed with TM and scheduled for follow-up examinations. At age 4, her VA was 20/25 in both eyes and there was no progression of the lesion.

Case 2– A 8-year-old Caucasian girl was referred to our retina clinic for a macular lesion detected in her left eye. Her VA was 20/20 bilaterally. Her right fundus was unremarkable; however, there was a well-circumscribed, hypopigmented, and torpedo-shaped lesion in the left temporal macula, whose tip was pointing towards the fovea (Fig. 2a). Optical

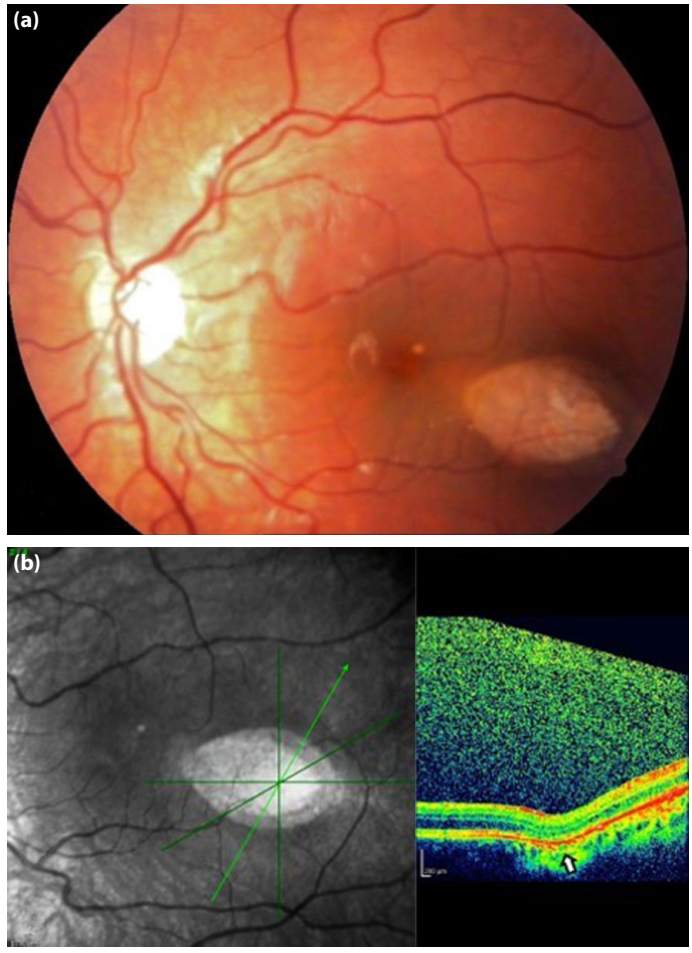

Fig. 2. **(a)** Color fundus photo of the left eye shows a well-circumscribed, hypopigmented, and torpedo-shaped lesion in the left temporal macula. **(b)** Disruption and thinning of outer retina with increased choroidal reflectivity in the region and a focal choroidal excavation (arrow) are prominent in optical coherence tomography.

coherence tomography (OCT) scan of the lesion revealed a thin outer retina with increased choroidal reflectivity under the lesion with a prominent focal choroidal excavation (Fig. 2b). There was no associated subretinal cleft.

Case 3– A 61-year-old Caucasian woman was consulted for a right macular lesion. Her VA's were 20/20 in both eyes. While the left fundus was normal, there was a torpedo-shaped hypopigmented lesion with a slight pigmentation at its temporal tail in the right eye (Fig. 3a). OCT of the lesion revealed a large subretinal cleft with a choroidal excavation. The upper roof of cleft had a brushy appearance (Fig. 3b). A well-defined window defect-like lesion exhibiting some staining was noted with fluorescein angiography (FA) (Fig. 3c). The lesion was apparent on the outer retina and choriocapillaris slabs on optical coherence tomography angiography (OCTA) (Fig. 4).

Verbal informed consent was obtained in all cases.

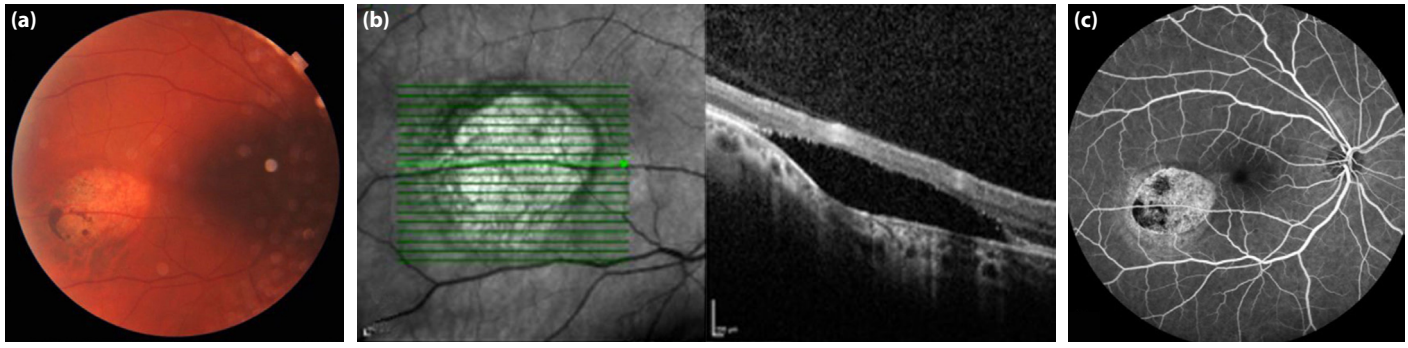

Fig. 3. **(a)** Color fundus photo of right eye demonstrates a torpedo-shaped hypopigmented lesion with segmental hyperpigmentation to its temporal edge. **(b)** Optical coherence tomography shows a large subretinal cleft and outer retinal excavation with thinning of outer retinal layers. **(c)** Fundus fluorescein angiography reveals a well-defined hyperfluorescent lesion with temporal hypofluorescence corresponding to hyperpigmented areas of torpedo lesion. Lesion shows no leakage in late-phase frames.

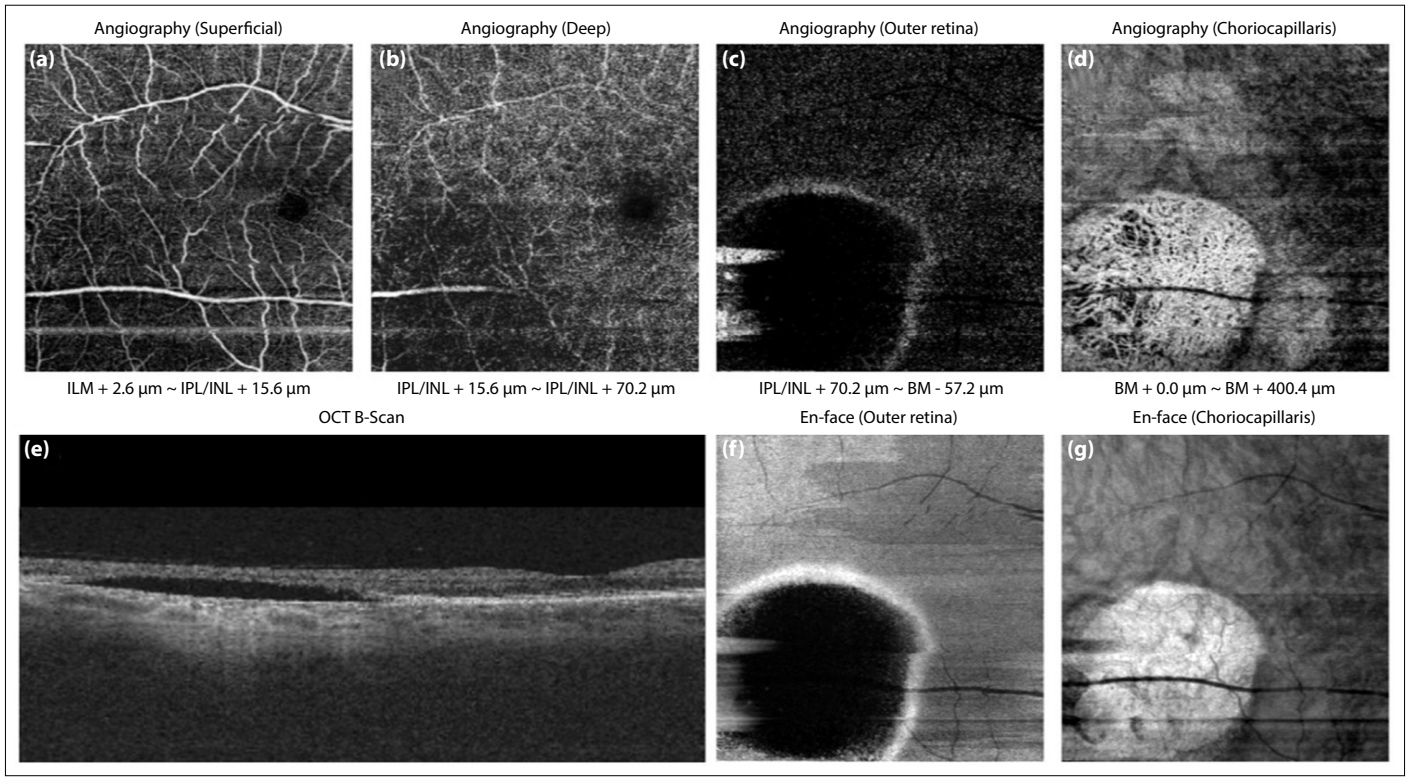

Fig. 4. **(a-d)** Optical coherence tomography angiography reveals normal superficial and deep plexi, and a convoluted pattern of fine vessels with some hyporeflective spaces between them on the choriocapillaris slab. **(e)** A subretinal cleft is prominent in swept-source optical coherence tomography (OCT). **(f-g)** En face OCT slabs of outer retinal layers and choroid show a homogeneous hyporeflective area corresponding to the subretinal cleft.

Discussion

TM is usually diagnosed incidentally during the ophthalmic examination as it is almost often asymptomatic. Patients can be at any age at the time of diagnosis (range, 6 months–72 years) and there is no predilection for a particular gender or race.[6,7] The youngest of our cases was 2 years old, whereas the oldest was 61. Although bilateral cases are reported, most of the TM lesions are typically unilateral. So far, all reported TM lesions were temporal to the macula, with one exception presenting with a nasal lesion.[8] In our case series, all TM lesions were unilateral and located temporally to macula. Although known as a benign lesion, it has been rarely associated with MNV and visual prognosis was good with intravitreal anti-VEGF injections according to case reports.[5,7] MNV was not observed in any of our cases.

Multimodal imaging has provided a better understanding of the structure of TM lesions. The characteristic OCT finding is an increase in choroidal reflectivity, with or without RPE hy-

perreflectivity, seen in the presence of normal inner retinal layers. The outer retina may be irregular due to RPE thinning and photoreceptor loss, and subretinal cleft may be present. Wong et al.[9] categorized TM according to the OCT findings into two subtypes. Having increased signal transmission to choroid and normal inner retinal structure in common, type 1 TM included the lesions without outer retinal cavitation, whereas type 2 TM included the lesions with outer retinal cavitation, which may be associated with inner choroidal excavation. Tripathy et al.[10] provided the most up-to-date classification of TM by defining a third type in which focal choroidal excavation is seen without cavitation. According to these definitions, while the OCT findings of Patient 1 and Patient 3 were compatible with type 2 TM, the lesion of Patient 2 could be classified as type 3 TM. Papastefanou et al.[11] described the characteristics of torpedo lesions with OCTA and observed hyporeflectivity at choriocapillaris level, indicating the atrophy, which in turn showed a correlation with subretinal cleft area on the OCT. In accordance with this description, there was a convoluted pattern of fine vessels with some hyporeflective spaces in between them in the choriocapillaris layer, while superficial and deep retinal layers appeared normal on the OCTA examination of Patient 3. Moreover, OCTA may reveal the increased density of choroidal vasculature due to increased signal transduction of thin RPE in cases of TM without subretinal cleft.[12] In most cases, FA revealed a well-defined hyperfluorescent window defect-like stained lesion without any leakage that probably indicates a loss of functional RPE. To date, no histopathologic evidence exists to confirm the true etiology of TM.

Differential diagnosis of TM includes other pigment-related lesions such as inactive toxoplasma retinochoroiditis, amelanotic nevus and/or melanoma of choroid, solitary congenital hypertrophy of RPE, and congenital RPE lesions having the potential of malignancy as in Gardner's syndrome.[7] Therefore, it is of utmost importance for the physician to recognize the lesion correctly.

Conclusion

TM usually presents as a hypopigmented "torpedo-like" lesion temporal to macula. Although retinal thinning, structural changes in the outer retinal layers and choriocapillaris atrophy are observed with multimodal imaging, VA is typically not affected. In some cases, it has been associated with conditions requiring treatment such as MNV. Various retinochoroidal lesions should be considered in its differential diagnosis, including tumors with systemic involvement. With this report, we want to share our observations on TM and to remind the ophthalmologists about the entity.

Informed Consent: Written informed consent was obtained from the parents and the case for the publication of this case report and accompanying images.

Peer-review: Externally peer-reviewed.

Authorship Contributions: Concept: A.O.S.; Design: A.O.S.;; Supervision: A.O.S.; Data Collection and/or Processing: A.T.B., A.O.S., Z.A.; Analysis and/or Interpretation:A.T.B., A.O.S., Z.A.; Literature Search: C.D.E.; Writing: C.D.E., A.O.S.; Critical Reviews: A.O.S.

Conflict of Interest: None declared.

Financial Disclosure: The authors declared that this study received no financial support.

References

1. Roseman RL, Gass JD. Solitary hypopigmented nevus of the retinal pigment epithelium in the macula. Arch Ophthalmol 1992;110:1358–9. [CrossRef]
2. Daily M. Torpedo maculopathy or paramacular spot syndrome. In: New Dimensions in Retina Symposium; 1993.
3. Shields CL, Guzman JM, Shapiro MJ, Fogel LE, Shields JA. Torpedo maculopathy at the site of the fetal "bulge". Arch Ophthalmol 2010;128:499–501. [CrossRef]
4. Chawla R, Pujari A, Rakheja V, Kumar A. Torpedo maculopathy: A primary choroidal capillary abnormality? Indian J Ophthalmol 2018;66:328–9.
5. Shirley K, O'Neill M, Gamble R, Ramsey A, McLoone E. Torpedo maculopathy: Disease spectrum and associated choroidal neovascularisation in a paediatric population. Eye (Lond) 2018;32:1315–20. [CrossRef]
6. Kara C, Petriçli İS. Torpedo maculopathy in a 6-month-old infant: Early clinical and optical coherence tomography findings. J Pediatr Ophthalmol Strabismus 2017;54:e54–7. [CrossRef]
7. Williams PJ, Salek S, Prinzi RA, Bergstrom C, Hubbard GB. Distribution patterns of torpedo maculopathy: Further evidence of a congenital retinal nerve fiber layer-driven etiology. Saudi J Ophthalmol 2019;33:260–7. [CrossRef]
8. Buzzonetti L, Petroni S, Catena G, Iarossi G. Optical coherence tomography and electrophysiological findings in torpedo maculopathy. Doc Ophthalmol 2015;130:65–70. [CrossRef]
9. Wong EN, Fraser-Bell S, Hunyor AP, Chen FK. Novel optical coherence tomography classification of torpedo maculopathy. Clin Exp Ophthalmol 2015;43:342–8. [CrossRef]
10. Tripathy K, Sarma B, Mazumdar S. Commentary: Inner retinal excavation in torpedo maculopathy and proposed type 3 lesions in optical coherence tomography. Indian J Ophthalmol 2018;66:1213–4. [CrossRef]
11. Papastefanou VP, Vázquez-Alfageme C, Keane PA, Sagoo MS. Multimodality imaging of torpedo maculopathy with swept-source, en face optical coherence tomography and optical coherence tomography angiography. Retin Cases Brief Rep 2018;12:153–7. [CrossRef]
12. Menezes K, Mancera N, Patel H, Kattih Z, Mhaskar R. Torpedo maculopathy: A systematic review of case reports. Ophthalmic Surg Lasers Imaging Retina 2021;52:78–83. [CrossRef]