

Torpedo Maculopathy in Multimodal Imaging: New Perspectives in Light of Diagnosis

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ABSTRACT

Purpose: To present a pediatric case of Type 2 torpedo maculopathy with distinct choriocapillaris flow reduction demonstrated by optical coherence tomography angiography (OCT-A), highlighting the potential prognostic value of vascular assessment in this condition.

Case Presentation: A 7-year-old girl was found to have a well-defined hypopigmented lesion in the inferotemporal macular region during routine examination. Visual acuity and anterior segment findings were normal. Spectral-domain optical coherence tomography (SD-OCT) revealed outer retinal thinning, ellipsoid zone disruption, and focal choroidal excavation, consistent with Type 2 torpedo maculopathy. OCT-A demonstrated reduced vascular density at the choriocapillaris level, while fluorescein angiography and fundus autofluorescence showed changes compatible with retinal pigment epithelium alteration.

Conclusion: This case underscores that torpedo maculopathy, particularly Type 2 forms in pediatric patients, may involve microvascular alterations detectable with OCT-A. These findings suggest that the condition may not always be structurally static and that periodic OCT-A follow-up may be warranted even in asymptomatic individuals.

Keywords: Torpedo maculopathy, multimodal imaging, optical coherence tomography (OCT), optical coherence tomography angiography (OCT-A)

INTRODUCTION

Torpedo maculopathy (TM) is a rare congenital anomaly of the retinal pigment epithelium (RPE) that is generally considered benign (1). It typically manifests as a well-demarcated, torpedo-shaped lesion in the macular region, pointing toward the fovea. TM is often detected incidentally during routine ophthalmoscopic examinations and rarely causes significant visual symptoms. However, developmental anomalies in the retinal layers may be observed (2).

Multimodal imaging techniques play a crucial role in confirming the diagnosis. Fundus photography provides an overview of the lesion's morphology, while FAF imaging

offers valuable information regarding RPE integrity. The presence of hyperautofluorescent borders surrounding the lesion is considered one of the characteristic features of TM (3). SD-OCT enables detailed examination of retinal layers (4), and OCT-A allows for an in-depth assessment of retinal and choroidal vascular networks (5). Additionally, FA reveals window defects caused by RPE atrophy, leading to increased fluorescence permeability (6).

This comprehensive imaging approach facilitates the detailed characterization of TM's structural and functional abnormalities. It also serves as an essential reference for monitoring disease progression and developing treatment

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strategies, particularly in young patients, where TM subtypes may vary and require consideration during diagnosis and follow-up (3,4).

The primary objective of this study is to highlight the critical role of multimodal imaging techniques in diagnosing TM. These advanced imaging methods enable a precise evaluation of the lesion's structural and functional properties, thereby optimizing diagnostic accuracy and patient management. Our case study aims to contribute to the development of more effective and personalized approaches in diagnosis and follow-up.

CASE REPORT

A seven-year-old female patient presented with complaints of blurred vision in the right eye. Visual acuity assessment with the Snellen chart revealed full visual acuity in both eyes. Intraocular pressure measurements were within normal limits, and the anterior segment examination was unremarkable.

Fundus examination revealed a well-defined hypopigmented excavation area in the inferotemporal macular region of the right eye, with normal optic disc morphology. To confirm the diagnosis, the patient underwent fundus photography, OCT, OCT-A, FA, FAF, and infrared (IR) imaging. (Table 1)

Fundus photography revealed an excavation area in the inferotemporal macula (Figure 1A). SD-OCT demonstrated thinning of the outer retina and photoreceptor loss at the lesion site (Figure 1B). OCT-A evaluation showed no significant thinning in the superficial and deep retinal layers, but thinning of the choroidal vascular network and reduced vascular density at the choriocapillaris level were observed (Figure 1C). FA revealed an early hyperfluorescent area due to window defects (Figure 1D). FAF imaging highlighted hypoautofluorescence within the lesion (Figure 1E). IR imaging provided clearer visualization of lesion boundaries (Figure 1F). Based on multimodal imaging findings, a diagnosis of torpedo maculopathy was established.

Table 1. Multimodal Imaging Characteristics and Clinical Interpretation in Torpedo Maculopathy

Imaging Modality	Findings	Clinical Interpretation
Fundus Photography	Well-defined torpedo-shaped hypopigmented lesion inferotemporal to the fovea	Characteristic morphology of torpedo maculopathy
SD-OCT	Outer retinal thinning, ellipsoid zone disruption, focal choroidal excavation	Consistent with Type 2 torpedo maculopathy
OCT-A	Reduced vascular density at the choriocapillaris level	Indicates vascular involvement and may have prognostic significance
FA	Early hyperfluorescence due to window defect, no leakage	Confirms RPE atrophy without neovascularization
FAF	Hypoautofluorescent core with subtle hyperautofluorescent border	Reflects RPE alteration and metabolic stress
IR Imaging	Clear delineation of lesion borders	Enhances visualization of lesion contour and extent

SD-OCT: Spectral-domain optical coherence tomography; OCT-A: Optical coherence tomography angiography; FA: Fluorescein angiography; FAF: Fundus autofluorescence; IR: Infrared imaging; RPE: Retinal pigment epithelium.

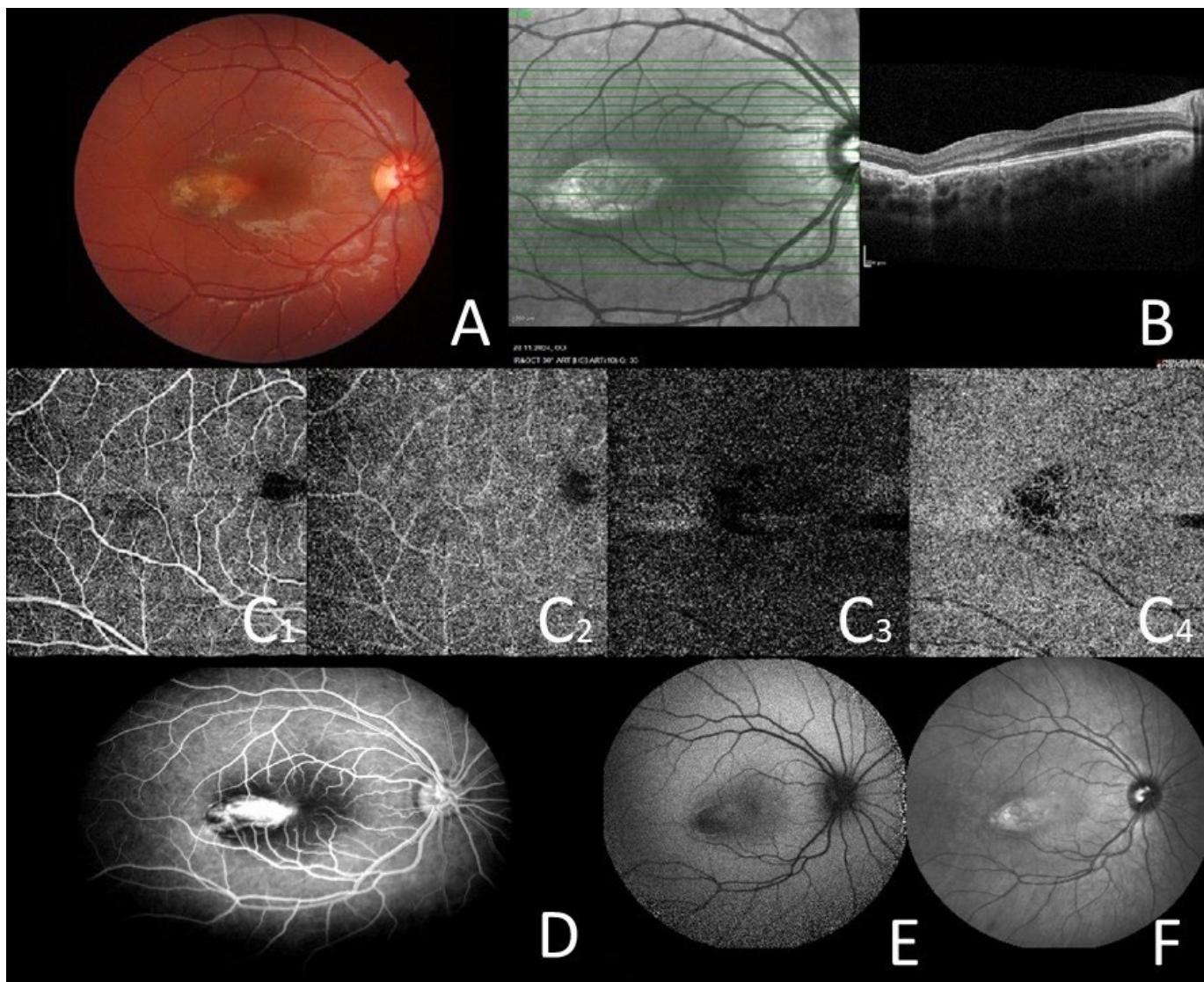


Figure 1. Multimodal imaging findings in torpedo maculopathy.

(A) Fundus photograph showing a well-demarcated hypopigmented lesion in the inferotemporal macula.

(B) Spectral-domain optical coherence tomography (SD-OCT) revealing outer retinal thinning, photoreceptor loss, and disruption of the retinal pigment epithelium (RPE) at the lesion site.

(C₁-C₄) Optical coherence tomography angiography (OCT-A) images demonstrating normal superficial and deep retinal layers, with decreased vascular density at the choriocapillaris level and choroidal vascular network attenuation.

(D) Fluorescein angiography (FA) depicting early hyperfluorescence due to window defects associated with RPE atrophy.

(E) Fundus autofluorescence (FAF) imaging highlighting hypoautofluorescent regions corresponding to the lesion.

(F) Infrared (IR) imaging providing enhanced delineation of lesion boundaries.

DISCUSSION

Torpedo maculopathy is a congenital anomaly of the RPE, characterized by a torpedo-shaped lesion directed toward the fovea (1). While conventional fundus photography outlines the lesion's morphology, it does not provide a complete structural and functional assessment. Therefore, multimodal imaging approaches are crucial for a comprehensive evaluation (3).

Structurally, SD-OCT findings are essential for differentiating torpedo maculopathy subtypes.

SD-OCT has demonstrated thinning of the outer retina, ellipsoid zone loss, and interdigitation zone disruption in TM lesions (4). These findings support the hypothesis that TM arises from a developmental defect leading to abnormalities in RPE morphology (1). Additionally, subretinal cavitation or choroidal excavation variations contribute to the classification of different TM subtypes (4). Wong et al. proposed two morphological subtypes of torpedo maculopathy based on SD-OCT characteristics. Type 1 lesions are characterized by preservation of the outer retinal layers with minimal structural alteration, whereas Type 2 lesions demonstrate ellipsoid zone disruption, outer retinal thinning, and focal choroidal excavation (7). In our case, the presence of distinct photoreceptor layer attenuation and focal excavation on SD-OCT corresponds to Type 2 torpedo maculopathy. This distinction is clinically relevant, as Type 2 lesions may indicate a greater degree of retinal and choroidal involvement, supporting the need for more careful long-term monitoring, even in asymptomatic pediatric patients.

FAF imaging highlights hyperautofluorescent borders around the lesion, reflecting RPE dysfunction (7). FA supports TM diagnosis by revealing window defects, which indicate increased fluorescence permeability due to RPE atrophy (6). These techniques provide insights into both structural abnormalities and RPE functionality.

In addition to structural alterations, vascular involvement can be evaluated using OCT-A.

OCT-A facilitates detailed visualization of retinal and choroidal vasculature, which is particularly useful for ruling out complications such as choroidal neovascularization (7).

In our case, OCT-A revealed a distinct reduction in vascular density at the choriocapillaris level, suggesting that torpedo maculopathy may involve not only structural alterations of the outer retina but also functional compromise of the choroidal microvasculature. This finding aligns with the Type 2 morphological pattern described by Wong et al., in which outer retinal attenuation and focal choroidal excavation are present (7). This finding may have prognostic relevance, supporting the need for periodic OCT-A monitoring even in asymptomatic pediatric cases.

A recent systematic review including 110 published cases reported that torpedo maculopathy is generally regarded as a benign, nonprogressive, and predominantly unilateral lesion, typically identified incidentally during routine examination (8). The authors emphasized that SD-OCT most commonly demonstrates outer retinal layer thinning and RPE irregularity, with fluorescein angiography findings being generally non-specific. However, most cases in the literature did not include detailed vascular assessment with OCT-A, and therefore the potential role of choriocapillaris flow alterations remains insufficiently characterized (8).

These structural and vascular characteristics have practical implications for patient follow-up.

In our case, we utilized the aforementioned imaging methods, and our findings aligned with existing literature, confirming the diagnosis of type 2 torpedo maculopathy.

The integrated use of multimodal imaging techniques significantly enhances diagnostic accuracy by providing a detailed characterization of the structural and functional aspects of torpedo maculopathy. These methods are not only critical for assessing the current state of the disease but also serve as key references for long-term monitoring and early intervention. As technological advancements continue, diagnosing TM will become increasingly accurate and efficient.

This case highlights that choriocapillaris flow impairment can be detected at a very early age in Type 2 torpedo maculopathy, suggesting that the condition may involve progressive microvascular alteration rather than representing a purely static congenital lesion.

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