

## Brief Report

## Morpho-functional evaluation of torpedo maculopathy with optical coherence tomography angiography and microperimetry

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## ABSTRACT

**Purpose:** To report the case of a 13-year-old girl with torpedo maculopathy, evaluated with multimodal morpho-functional retinal imaging, including fundus photography, infra-red and blue fundus autofluorescence, swept-source optical coherence tomography (OCT), en face OCT, OCT angiography and microperimetry (MP).

**Observations:** On fundus examination, a torpedo-like hypopigmented lesion was observed temporal to the fovea in the left eye. OCT showed disruption of outer retinal layers and the presence of a subretinal cleft. On OCTA, a diffuse attenuation of signal from choriocapillaris was observed along the lesion. Functional analysis with MP revealed a reduction of retinal sensitivity over the lesion.

**Conclusions:** and importance: On OCTA, torpedo maculopathy is characterized by vascular alterations of the choriocapillaris along the lesion.

## 1. Introduction

Torpedo maculopathy is a rare retinal unilateral anomaly first reported by Gass and Roseman in 1992.<sup>1</sup> The lesion is currently considered the result of presumably congenital retinal pigment epithelium (RPE) abnormalities, with a variable degree of disruption of external retinal layers.<sup>2–4</sup> Despite its unique shape and location, this condition presents different qualitative features on retinal imaging, with variable functional alterations throughout the lesion. Although the torpedo lesion has been carefully analyzed with optical coherence tomography (OCT) and fluorescein angiography, up to date only very few reports exist regarding its evaluation with optical coherence tomography angiography (OCTA).<sup>5–7</sup> Thus, additional data are needed to better understand which multilayer microvasculature alterations are associated with torpedo lesions.

In this study, the authors report a case of torpedo maculopathy evaluated with multimodal morpho-functional retinal imaging, including OCTA and microperimetry (MP).

## 2. Case report

An asymptomatic 13-year-old girl with an incidentally found macular lesion in her left eye was referred to our institution for further evaluation.

The patient underwent complete ophthalmic examination with

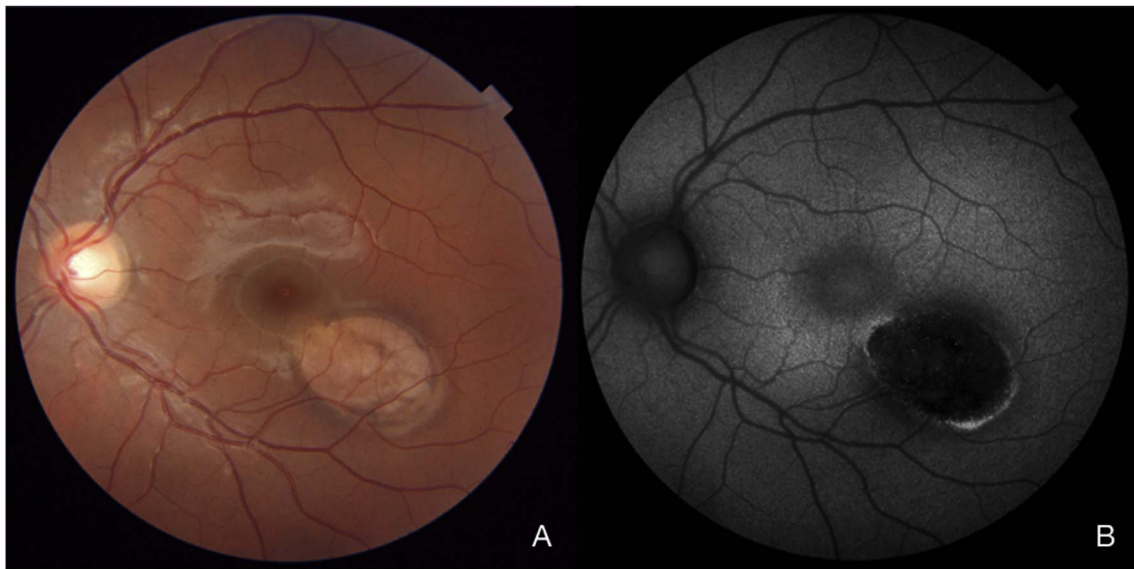
measurement of best corrected visual acuity and multimodal fundus imaging, including fundus photography, infra-red and fundus autofluorescence (FAF) imaging, swept-source optical coherence tomography (SS-OCT) and OCT angiography (DRI OCT Triton, Topcon, Tokyo, Japan), and microperimetry (MP1 Microperimeter; Nidek, Gamagori, Japan). Best corrected visual acuity was 20/20 in both eyes and ophthalmic, medical, and family histories were unremarkable.

On fundus examination, a flat torpedo-like hypopigmented lesion of 3.4 mm × 2.4 mm was observed infero-temporal to the fovea in the left eye, with loss of autofluorescence throughout the lesion on FAF (Fig. 1). SS-OCT showed mild outer retinal cavitation, with thinning of outer nuclear layer and disruption of the myoid, ellipsoid and interdigitation zones (Fig. 2). A large subretinal cleft was observed in correspondence of the lesion, associated with irregularity and thinning of RPE-Bruch complex. Mild increased signal transmission in the choroid was highlighted below the cleft, with no clear alterations of Sattler and Haller layers.

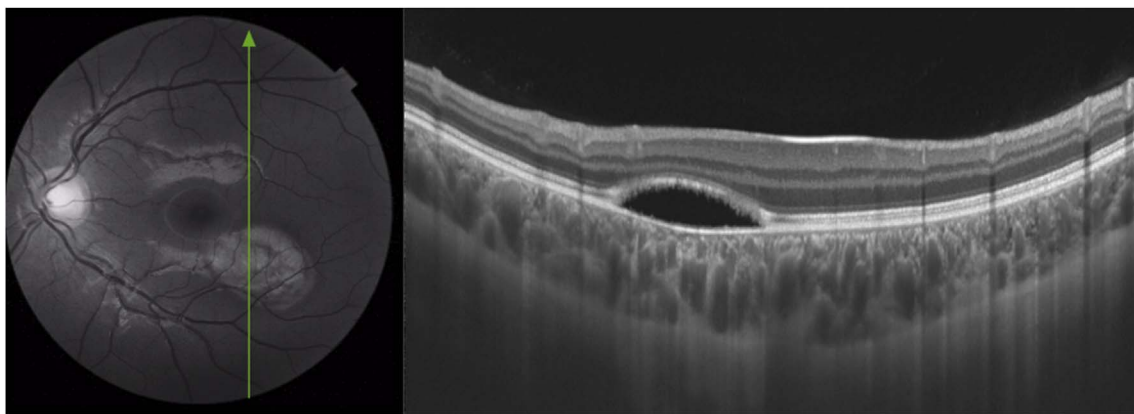
OCTA showed no alteration of superficial capillary plexus, with only focal areas of loss in the deep capillary plexus (Fig. 3). Conversely, a diffuse homogenous attenuation of signal from the choriocapillaris was observed within the lesion (Fig. 3-D). Choroidal vessels appeared normal, with *angiographic* findings similar to the surrounding unaffected tissue.

*En face* OCT focused on external retinal layers revealed a homogeneous hyporeflective area corresponding to the subretinal cleft

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**Fig. 1. Fundus imaging of torpedo maculopathy.** (A) Fundus photography showing torpedo-like lesion located infero-temporal to the fovea in the left eye. The lesion appears largely hypopigmented with focal peripheral areas of hyper-pigmentation. (B) On fundus autofluorescence, the lesion shows a homogeneous loss of autofluorescence, with a hyperfluorescent halo surrounding the torpedo lesion.



**Fig. 2. Optical coherence tomography features of torpedo maculopathy.** Swept source optical coherence tomography showed mild outer retinal cavitation, with thinning of outer nuclear layer and disruption of the myoid, ellipsoid and interdigitation zones. Along the lesion, a large subretinal cleft was observed, associated with irregularity and thinning of the RPE-Bruch complex and a mild increase of signal transmission in the choroid below the cleft, with no clear alterations of Sattler and Haller layers.

(Fig. 3-F).

Functional analysis with MP revealed a variable reduction of retinal sensitivity throughout the lesion, with a mean retinal sensitivity of 11.26 dB (Fig. 4).

At 9 months of follow-up, the morpho-functional features of the lesion were unchanged.

### 3. Discussion

Since its first description in 1992<sup>1</sup>, torpedo maculopathy has been sporadically reported in a limited number of case reports and small case series. Currently considered a rare congenital condition, it is characterized by a unilateral hypopigmented torpedo-shaped lesion in the temporal macula, with the tip pointing to the fovea and a variably pigmented tail.<sup>2</sup> Although its pathogenesis is still unknown, several hypotheses have been proposed: a developmental defect in the nerve fiber layer along the horizontal raphe, abnormal choroidal or ciliary vasculature development and, finally, a persistent developmental defect in the RPE in the fetal temporal bulge.<sup>2,8</sup>

Extensively analyzed with OCT, torpedo maculopathy has been recently classified in two distinct types according to the pattern of

abnormality: type 1, with attenuation of outer retinal structures without outer retinal cavitation and type 2, showing both attenuation and cavitation of the outer retina.<sup>3</sup>

OCT-A is an innovative imaging tool allowing the non-invasive study of retinal and choroidal vasculature through the acquisition of volumetric angiographic images, with great potential for the evaluation of a wide range of retinal conditions in the clinical setting.<sup>9</sup>

To the best of our knowledge, only three case series have been published in the literature describing the OCT-A features of torpedo lesions.<sup>5–7</sup>

In a recent small case series of two patients with torpedo maculopathy, evaluated with OCTA, Papastefanou et al.<sup>5</sup> reported a normal superficial retinal plexus and attenuation of the deep retinal vasculature along the lesion, associated with loss of deep vessels in correspondence of the subretinal cleft. These reported findings are dissimilar from our case, where only minute isolated modifications of the deep retinal vascular network were observed. Considering the younger age of our patient, it could be supposed that alterations of deep retinal layers, including changes to retinal vascular plexus, gradually develop with time and would not represent the primary site of torpedo maculopathy.

If we consider the modifications of the choroid, a diffuse attenuation

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