Torpedo maculopathy—inferior variant

Clinical Presentation

A 19-year-old man was referred to the retina specialist for an incidental finding of a right hypopigmented choroidal lesion. The patient was asymptomatic and had no significant past ocular or medical history. Visual acuity was 20/20 bilaterally. Intraocular pressure was normal. Slit lamp examination was unremarkable bilaterally, and the vitreous cavity was clear. Fundus examination showed a healthy optic nerve appearance with a cup-to-disc ratio of 0.5 in both eyes. Examination of the right posterior pole revealed a torpedo-shaped hypopigmented lesion in the inferior macula, with the nasal apex pointing towards the optic disc (Figure 1A). Fundus examination of the left eye was unremarkable.

Discussion

The term "torpedo maculopathy" was first coined by Daily in 1993 to describe a characteristic torpedo-shaped lesion in the temporal macula.[1] Since then, multiple reports of similar lesions have been added to the literature that has contributed to better characterization of this entity.

The classic torpedo maculopathy lesion is characterized as an oval-shaped hypopigmented lesion in the temporal macula pointing towards the fovea. The lesion can have relative hyperpigmentation of the borders, especially the temporal aspect, but never on the nasal border. Choroidal vessels are not visible over the lesion, unlike in chorioretinal atrophy. The borders of the lesion form a regular oval shape with a sharply demarcated nasal border. Vision is almost always unaffected, but there can be visual field defects corresponding to the lesion on testing. Autofluorescence imaging reveals uniform hypoautofluorescence over the lesion (Figure 1B). There may be a border of hyperautofluorescence corresponding to lipofuscin accumulation seen often inferiorly. Fluorescein angiography reveals a hyperfluorescent window defect over the lesion without leakage. Optical coherence tomography (OCT) of the lesion reveals intact inner retinal layers, often attenuated outer retina with photoreceptor loss (Figure 2). There may be outer retinal cavitation where there is photoreceptor loss without neurosensory elevation, or subretinal fluid with neurosensory elevation, which is presumably from functional impairment of the retinal pigment epithelium (RPE).[2] There is both RPE and choroidal hyperreflectivity. Inner choroidal excavation has also been described in some cases.[3] The main differentiating feature from a congenital hypertrophy of the RPE (CHRPE) is that with CHRPE, there is RPE thickening and decreased hyperreflectivity of the choroid on OCT.

Early presentation in infancy and long-term stability over time, as well as the consistent localization of cases to the temporal macula support a congenital etiology with a conserved mechanism; however, the exact pathogenesis is yet to be agreed upon.

Several theories have been proposed including abnormal depigmented melanin content in RPE cells, disruption of RPE differentiation owing to a disturbance in choroidal vasculature, a defect in RPE development at the site of the fetal temporal bulge, and incomplete differentiation of the nerve fibre layer and RPE.[4–6] In our case, apart from mild photoreceptor atrophy, there is no nerve fibre layer, RPE, or retinal defect overlying the lesion.

Although the majority of torpedo maculopathy cases are located along the location of the horizontal raphe, our case is unique in that the lesion is located in the inferior macula

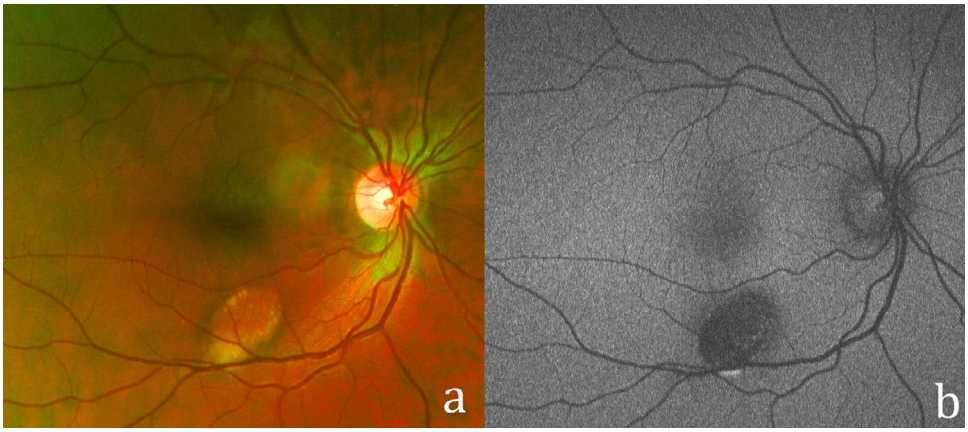

Fig. 1—Color fundus photo of the right eye showed inferior torpedo maculopathy, with the lesion pointing towards the optic nerve (A). Fundus autofluorescence showed hypoautofluorescence corresponding to the torpedo lesion with a hyperautofluorescent lower edge (B).

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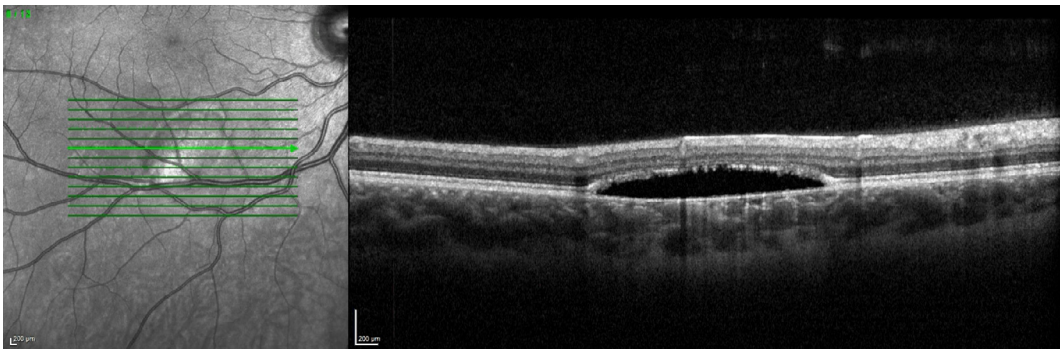

Fig. 2—Optical coherence tomography images of the lesion revealed preserved inner retinal layers and neurosensory retinal elevation with subretinal fluid.

with the nasal tip pointing towards the optic nerve. To our knowledge there is only one other case of torpedo maculopathy reported outside the temporal raphe location, also situated inferiorly along the nerve fibre layer pointing to the optic disc.[7]

This case highlights that torpedo maculopathy can rarely be located outside the horizontal raphe, in this case in the inferior macula, but still orientated along the nerve fibre layer path with the nasal apex pointing toward the optic disc.

Supplementary materials

Supplementary material associated with this article can be found in the online version at doi:10.1016/j.jcjo.2021.02.008.

Meghan J. Smith, David I.T. Sia, Mark Greve
Faculty of Medicine & Dentistry, University of Alberta, Edmonton, Alb.

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Correspondence to:
Mark Greve, MD: mgreve@ualberta.ca.

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Footnotes and Disclosure

The authors have no proprietary or commercial interest in any materials discussed in this article.