THE **RETINA** PARTNERS

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Case of the Month – April 2019

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A 42 year-old asymptomatic female was referred for an incidentally noted macular scar in the right eye. Visual acuity was 20/20 in both eyes. Intraocular pressures were within normal limits. The anterior segment examination was unremarkable, with absence of signs of present or past inflammation. Fundus examination of the left eye was unremarkable. Imaging of the right fundus is shown below.

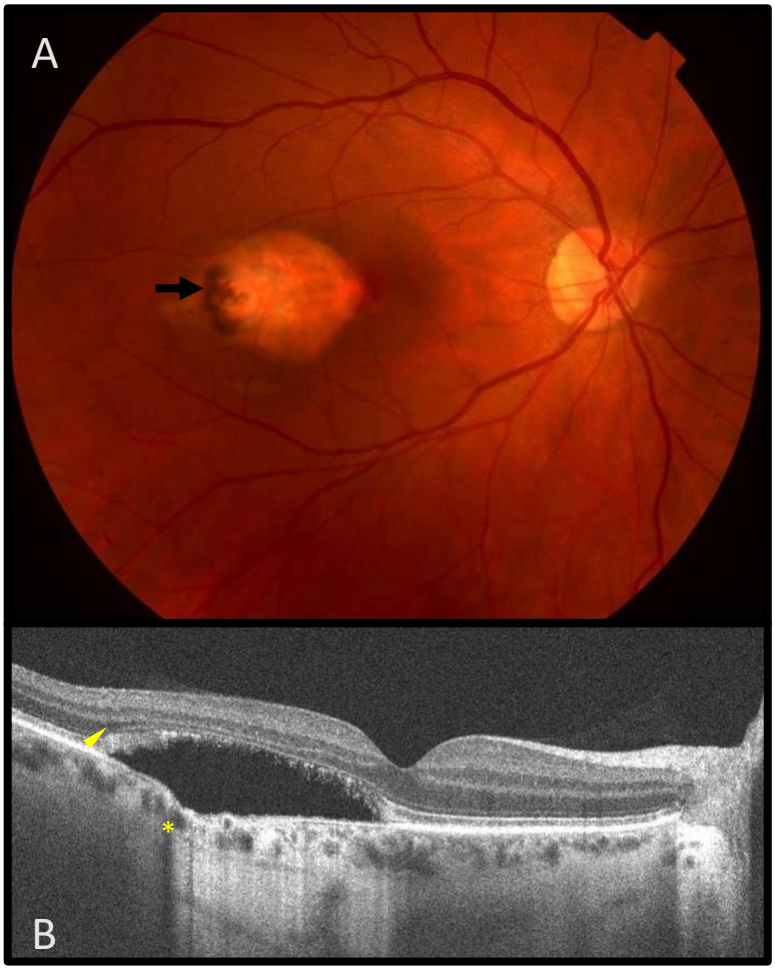

Figure 1: A. Color fundus photo of the right eye shows an ovoid, hypopigmented lesion in the temporal macula with hyperpigmentation at its temporal edge (arrow). **B.** SD-OCT through the lesion shows a focal excavation of the choroid with adjacent hyper-transmission (asterisk). There is an overlying outer retinal cavitation with elevation of the neurosensory retina, and thinning of the outer nuclear layer (arrowhead)**.**

Differential Diagnosis: Amelanotic choroidal nevus, chronic central serous retinopathy, torpedo maculopathy, toxoplasmosis chorioretinopathy, choroidal osteoma, choroidal hemangioma, chorioretinal scar,

Clinical Course:

The patient was diagnosed with torpedo maculopathy on the basis of imaging and exam findings. She has remained stable, with 20/20 vision and no visual complaints over the course of 7 years of follow up.

Discussion:

Torpedo maculopathy is a rare, congenital lesion of the retinal pigment epithelium in the macula, most commonly identified in asymptomatic children or young adults. Clinically, torpedo maculopathy appears as a hypopigmented, oval shaped and well defined lesion within the temporal macula. It takes its name from its characteristic pointed nasal tip aimed towards the fovea, making it appear like a torpedo. While the lesion is overall hypopigmented, the temporal border may have focal areas of hyperpigmentation (Figure 1A, arrow). The lesion may extend up to the foveal edge, but typically it does not involve the foveal center. While visual field testing may reveal a paracentral scotoma, patients are rarely symptomatic, and visual acuity is preserved.

Ancillary retinal imaging can play an important role in diagnosis and differentiation from other macular lesions. SD-OCT will show normal inner retinal structures, variable degrees of outer retinal disruption beginning with the outer nuclear layer, and attenuation of the RPE signal resulting in hyper-transmission into the choroid (figure 1B, asterisk). Two distinct subtypes of lesions have been identified on the basis of OCT. In type I lesions, the outer retinal structures display varying degrees of attenuation without a subretinal cavitation. In type II lesions, as in our case, there is an outer retinal cavitation with resultant elevation of the neurosensory retina (Figure 1B). Other imaging modalities like fluorescein angiography and fundus autofluorescence may also be helpful. Both show evidence of RPE loss within the lesion. Fluorescein angiography shows an early windowing defect. Importantly, there is no leakage on fluorescein testing, even in the type II lesions. This indicates the absence of a significant exudative component to the disease. Fundus autofluorescence will show a hypo-fluorescent signal, consistent with abnormal or absent RPE.

The pathogenesis of these congenital lesions remains uncertain, in part because no histopathologic studies exist to date. The majority of our knowledge comes from the imaging findings noted above, which indicate a focal loss of the retinal pigment epithelial cells and varying degrees of both focal choroidal and outer retinal disruption within the temporal macula. Some authors contend that this may be the result of abnormal choroidal or ciliary vascular development. Others have proposed a theory involving abnormal RPE development during embryogenesis, in an area termed the fetal temporal bulge. This is a region of temporarily high RPE cellular density just temporal to the fovea, which typically regresses by birth. Regardless of its pathogenesis, these lesions are benign, non-progressive and may be monitored without need for further intervention or testing.

Take Home Points

- Torpedo maculopathy presents with a characteristic location, shape and appearance.
- Torpedo maculopathy is a benign, and non-progressive congenital lesion which typically does not cause visual symptoms.
- Optical coherence tomography, fluorescein angiography and fundus autofluorescence can help distinguish this congenital condition from other macular lesions.

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