Two Cases of Torpedo Maculopathy with OCT scans, one demonstrating a unique deep pit with visible bare sclera.

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Abstract:

This case discussion presents two cases of torpedo maculopathy not previously diagnosed. One patient displays significant tissue disruption overlying the lesion, indicating possibility of full-thickness loss to neurosensory retina accompanying this rare retinal pathology.

Case Report:

A 27 year old white male visited the University Eye Care clinic complaining of longstanding blur in his right eye at both distance and near. He had a history of longstanding poor vision OD for “as long as he could remember,” and had been diagnosed with amblyopia as a child. He had been patched OD at the age of 5 for a period of about one year, although he noticed no change in vision after patching. He had no history of ocular surgery and no history of ocular trauma, although after questioning he mentioned a car accident in childhood. He could not recall any injury to eye, orbit or adnexa during the accident or at any other time. Family history was significant only for glaucoma (late onset, via his maternal grandmother). He suffered from occasional outbreaks of facial acne for which he was taking oral amoxicillin.

Best corrected visual acuities were: OD 20/50+2 , OS 20/20.

Extraocular motilities were full, pupils were equal, round, and reactive to light with no APD, and confrontation visual fields were full to finger counting in both right and left eyes. Cover tests revealed no tropia at distance or near (near cover test revealed mild exophoria).

Slit lamp examination revealed clear lids and lashes, intact and clear corneas of both eyes, flat and intact irides, deep and quiet anterior chamber with open angles via van Herick, and clear crystalline lenses. Intraocular pressure taken via Goldmann tonometry was 14 mm Hg in both eyes. Dilated fundus examination of the posterior pole revealed a deep, oval-shaped, well-circumscribed lesion immediately temporal to the fovea in the right eye, approximately ¾ DD in diameter. The edges of the lesion were flat and pigmented and the lesion appeared to extend through the neurosensory retina, RPE, and choroid, revealing bare sclera at its center. There was no vitreal traction at or around the lesion and funduscopy of the right eye revealed no other lesions or defects upon both examination of the posterior pole with 78D lens and examination of the periphery via binocular indirect ophthalmoscopy. Discs were pink with distinct margins and .1 C/D ratios in both eyes; the retinal vasculature of both eyes was healthy, and the macula of the left eye was flat and intact. The patient’s self-report of longstanding decreased vision OD indicated that the scar had most likely been present, and responsible for reduced VA OD, since childhood.

At follow-up examination, Amsler grid of the right eye revealed a discrete scotoma nasal to fixation with no areas of distortion. OCT imaging of the retina immediately adjacent to the center of the lesion
revealed areas of disorganization of the RPE surrounding a large excavated scar which extended through the neurosensory retina, pigment epithelium and choroid. The surrounding “ring” of disorganized RPE revealed a hyporeflective OCT signal and missing photoreceptor integrity line, and the neurosensory retina over the areas of hyporeflective OCT demonstrated attenuation as well as retinoschisis. No foveal pit was visible on OCT. The patient is to return to the Retina service of the University Eye Care Center for further testing, including Humphrey visual field to determine the extent of scotoma and fundus photography with autofluorescence.

A second patient, a 26 year old white male, presented to the Eye Institute at NOVA Southeastern University College of Optometry for a routine exam. He had no complaints as per history and systemic health was unremarkable, as was family history. He had no recollection of any ocular trauma or ocular surgery.

Entering visual acuity was 20/20, uncorrected, in each eye. Extraocular motilities were full, pupils were equal, round, and reactive to light, and confrontation visual fields were full to finger counting in both right and left eyes. Cover tests revealed no tropia at distance or near, and orthophoria at both distance and near.

Slit lamp examination revealed clear lids and lashes, intact and clear corneas of both eyes, flat and intact irides, deep and quiet anterior chamber with open angles via van Herick, and clear crystalline lenses of both eyes. Intraocular pressure taken via Goldmann tonometry was 15 mm Hg in both eyes. Dilated fundus examination of the posterior pole revealed an oval-shaped, well-circumscribed lesion temporal to the fovea, sparing the foveal pit, in the right eye, approximately 1.25 DD in diameter. The edges of the lesion were flat and pigmented. There was no vitreal traction at or around the lesion and fundoscopy of the right eye revealed no other lesions or defects upon both examination of the posterior pole with 78D lens and examination of the periphery via binocular indirect ophthalmoscopy. Discs were pink with distinct margins and .3 C/D ratios in both eyes; the retinal vasculature of both eyes was healthy, and the macula of the left eye was flat and intact.

**Differential diagnoses:**

The leading diagnosis in both cases is **torpedo maculopathy**, based on the characteristic shape and appearance of the lesions, the relatively good acuity, characteristic RPE hypoplasia at the edges of the lesion with overlying disorganization of neurosensory retina, the fact that it neatly bisects the horizontal raphe, and lack of history of trauma. Differentials are as follows:

**Toxoplasma retinochoroiditis** is a focal necrotizing chorioretinitis caused by the parasite *toxoplasma gondii*. Lesions are typically found in the posterior pole, with acute cases presenting as focal areas of chorioretinal inflammation accompanied by vitritis. Old, inactive lesions toxoplasmosis are relatively common and present as circular areas of hyperpigmented chorioretinal scarring. Acuity loss can be severe if the location of the chorioretinitis is foveal. A focal area of inflammation adjacent to an old chorioretinal scar is pathognomonic (4) as *toxoplasma gondii* cysts have a tendency to encircle old chorioretinal scars (3). Accompanying signs of acute, as opposed to congenital, toxoplasmosis include occlusive vasculitis, papillitis, granulomatous anterior uveitis or anterior uveitis resulting from anterior
“spillover” of posterior uveitis, vitritis, or CME (3). Reoccurrence of the infection can occur and suspected patients should be tested for toxoplasmosis antibody titre levels. The parasite may remain viable in cysts for over 25 years (4). In these patients, the longstanding nature of the lesion and the lack of acute inflammatory event would point to congenital, rather than acute, toxoplasma retinochoroiditis.

**Chorioretinal scarring secondary to choroidal rupture** is a possible complication of contusion injuries to the eye, resulting from the contrecoup shock waves generated in blunt trauma (3) Visible ruptures represent a break in Bruch’s membrane and the choroid. Ruptures are generally crescent-shaped as opposed to oval or circular, and CNV is a possible late-stage complication as a result to ruptures in Bruch’s membrane (3,4). Blunt trauma may also cause full-thickness macular holes via several mechanisms, including vitreous traction and contusion necrosis (4).

**Discussion: Torpedo maculopathy** has been described as a temporal parafoveal coloboma that is congenital in nature. The appearance is of a solitary temporal lesion with well-defined margins and a characteristic leading edge which points towards the macula (8) Researchers have theorized that its characteristic oval shape and location are due to incomplete differentiation of the arcuate bundles along the horizontal raphe (1). Other clinicians have described this finding as an amelenotic nevus of the retinal pigment epithelium (2). These lesions classically spare the fovea and in most cases cause no loss of acuity, although a scotoma is generally present corresponding to the location of the parafoveal coloboma that can be detected via visual field testing. (8) The lesions typically remain stable and do not involve risk of choroidal neovascularization (2).

While Gass describes this lesion as typically involving a focal loss of retinal pigment epithelium with histologically normal overlying retina, recent OCT studies of patients with torpedo maculopathy call this into question (2). Patients have been observed with overlying neurosensory retinal detachments, overlying photoreceptor thinning, and tissue loss. (2,6)

Interestingly enough, the histology of the retinal pigment epithelium as studied via OCT is also controversial in this clinical entity. In several case studies, Tsang et. al (7) found an RPE that was subtly hyperreflective and of normal thickness, yet with significant attenuation and disorganization of the inner and outer neurosensory retinal layers overlying the defect in RPE. In contrast, Golch et. al (8) describe a thin, abnormal RPE signal with overlying degenerated photoreceptors and loss of outer segments in several patients with torpedo maculopathy.

Tsang et. al point out that, because RPE is the first layer to differentiate before neural retina, colobomas affecting the RPE would be expected to involve both sensory retina and choroid because the differentiation of these tissues depends on an intact RPE layer (7). While choroidal colobomas can lead to visible bare sclera on fundus biomicroscopy, no previously reported case of torpedo maculopathy has presented with this finding, which is seen in our first patient with torpedo maculopathy. Classic chorioretinal colobomas arise infero-nasally from the optic disc, as opposed to temporal to the macula, and do not neatly bisect the horizontal raphe, as seen in our first patient.
Treatment:

Because torpedo maculopathy is non-progressive, no treatment is necessary. The patient was educated on the cause of longstanding decreased vision in his right eye, and given a full prescription with polycarbonate lenses. He is to return to the clinic for further testing and to be sent for toxoplasmosis antibody titre levels to ensure that he does not have a chorioretinal toxoplasmosis scar as was mentioned in the discussion section, with the possibility for re-occurrence of infection.

2) Yu Su, O.D., Andrew S. Gurwood O.D. Neurosensory retinal detachment secondary to torpedo maculopathy Optometry. Vol. 81 Issue 8 pp 405-407