

PEER REVIEWED

Torpedo Maculopathy

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Abstract

Torpedo maculopathy (TM) is a rare, congenital retinal lesion with a unique funduscopy presentation. TM lesions can be classified into two types based on their appearance on optical coherence tomography. This case report describes the diagnosis and management of a patient with type II TM. It also examines the epidemiology, pathophysiology, differential diagnosis and prognosis of the condition. TM is classically asymptomatic, benign and non-progressive and must be differentiated from other pathologies that are progressive or require intervention. Patient education for this condition is crucial to ensure awareness of the diagnosis and effective communication with doctors at future visits. Accurate diagnosis and classification of TM by an eyecare provider results in the most efficient and effective patient care.

Key Words: *torpedo maculopathy, retina, macula, optical coherence tomography*

Introduction

Torpedo maculopathy (TM) is a rare, congenital, predominantly unilateral macular lesion. TM lesions can be classified as type I or type II with the assistance of optical coherence tomography (OCT). Both classifications of the condition are typically non-progressive, but a few reports of accompanying progressive pathology exist in the literature.^{1,2} TM often presents with a classic fundoscopic appearance and a frank lack of symptomatology. Optometrists play a key role in properly diagnosing and managing this condition as it is most often first seen incidentally during a routine comprehensive eye exam. It is important to differentiate TM, a typically benign and non-progressive condition, from other progressive pathologies that may require treatment. Patient education is particularly important for rare conditions such as this because it allows patients to better communicate their diagnosis with future eyecare providers. This case report outlines the presentation, diagnosis and management of TM type II in a 68-year-old White man and is directed toward third- and fourth-year optometry students, optometry residents and practicing doctors.

Student Discussion Guide

Case presentation

A 68-year-old White male presented to the eye clinic for a comprehensive eye exam. His chief concern was bilateral near blur with his +2.75D over-the-counter reading glasses. He had no other associated symptoms such as eye strain or headaches at presentation. He was happy with his distance vision correction. According to previous records, the patient's ocular history was significant for mild non-proliferative diabetic retinopathy without macular edema in both eyes, dry eye syndrome from which he experienced relief with over-the-counter artificial tears, a bilateral blepharoplasty in 2015, hyperopia, presbyopia and a scar of unknown etiology adjacent to the macula in the left eye.

The patient's medical history was significant for abdominal pain, alcoholism, anxiety, cardiomyopathy, dermatitis, diabetes mellitus type II, diverticulitis, gastroesophageal reflux disease, hyperlipidemia, hypertension, ischemic heart disease, obstructive sleep apnea, surgery of back, knee and neck, and umbilical hernia. The patient's diabetes was managed with insulin, metformin and saxagliptin. His last blood sugar reading that morning was 118 mg/dL, and his most recent A1c was 8.0% one month prior. His other medications included aspirin, atorvastatin, baclofen, clopidogrel bisulfate, hydrochlorothiazide, lisinopril, metoprolol succinate, omeprazole, sertraline and spironolactone. The patient had no significant family ocular or medical history.

At presentation the patient's visual acuities with his distance correction were 20/25 OD and 20/20 OS. Lensometry readings of his distance correction were +1.75sph OD and +1.50sph OS. His entering near acuities with his over-the-counter reading glasses were 20/25 OD and OS. Pupils, extraocular muscle movements and confrontation fields were all unremarkable in both eyes. His vision was correctable to 20/20 OD and OS at distance and near with small changes in refraction, OD: +2.00sph, OS: +1.50sph, add: +1.50. The patient preferred a lower add for his age due to his tall stature and longer reading distance.



Figure 1. Fundus photographs of the right and left eye revealing mild non-proliferative diabetic retinopathy and a flat lesion temporal to the macula in the left eye consistent with torpedo maculopathy. [Click to enlarge](#)

Anterior segment evaluation was unremarkable in both eyes. Intraocular pressures by Goldmann applanation tonometry were 16 mmHg in each eye. The dilated ocular health exam revealed a few scattered hemorrhages and microaneurysms in the posterior pole of both eyes consistent with mild non-proliferative diabetic retinopathy. The macula was flat in both eyes without any apparent edema. The left eye had a lesion superior-temporal to the macula, beginning just adjacent to the fovea. The lesion appeared as a flat oval area of hypopigmentation with sharp borders and retinal pigment epithelium (RPE) hyperplasia at the temporal edge (**Figure 1**). The lesion was ellipsoid, longer horizontally at 1.5 disc diameters (DD) and 1 DD vertically. There was no evidence of edema, hemorrhaging or abnormal vasculature associated with the lesion. The optic nerve head appeared healthy and well-perfused and had a cup to disc ratio of 0.30/0.30 H/V in both eyes. There were no significant peripheral retinal findings in either eye.

The same day, fundus photography (Topcon TRC-NW200 Fundus Camera, Topcon Healthcare), OCT (Spectralis OCT, Heidelberg Engineering) and Humphrey visual field testing (Humphrey Visual Field Analyzer II-i, Carl Zeiss AG) were performed. The fundus photographs revealed mild non-proliferative diabetic retinopathy without macular edema in both eyes, and a unilateral macular lesion with a characteristic shape, appearance and location in the left eye (Figure 1). The OCT line scan through the lesion showed outer retinal attenuation and choroidal excavation consistent with type II TM (**Figure 2**). A 24-2 SITA-Fast test was performed in each eye. The visual field assessments were reliable in each eye. The field for the right eye was essentially clear, while the left eye had a dense central defect corresponding to the torpedo lesion location (**Figure 3**).



Figure 2. Optical coherence tomography single-line scan through the center of the macular lesion showed outer retinal attenuation and choroidal excavation consistent with torpedo maculopathy type II.¹² [Click to enlarge](#)



Figure 3. Humphrey visual field test results of the right and left eye showing an essentially normal field in the right eye and a dense to absolute defect in the left eye corresponding with the retinal lesion location. This pattern is consistent with torpedo maculopathy type II.¹² [Click to enlarge](#)

The patient was diagnosed with TM type II in the left eye, and the examination findings were explained to him. Because this condition is typically non-progressive, and the patient had good visual acuity, he was instructed to return in 1 year for his comprehensive eye examination. This recommended follow-up interval was also appropriate for his other examination finding of mild non-proliferative diabetic retinopathy without macular edema. At the next comprehensive eye exam 1 year later, the torpedo lesion and OCT findings were stable.

Educator's Guide

Key concepts

1. Recognize the signs and pattern of presentation of TM
2. Discuss differential diagnosis for TM and other similar retinal lesions
3. Review appropriate auxiliary testing to be performed as part of workup and management

Learning objectives

1. Describe the classic fundusoscopic presentation of TM and lack of symptoms
2. Describe other similar retinal lesions and how they differ from TM
3. Understand current theories of lesion epidemiology
4. Identify additional testing that should be performed to confirm diagnosis and further classify the lesion
5. Outline a management plan for a patient with TM

Discussion questions

1. Knowledge, understanding and facts about the clinical case and condition presentation
 - a. What is the pathophysiology of TM and how is it suspected that the lesion forms?
 - b. Describe the classic appearance and presentation of TM
 - c. Does a patient with TM typically present with related symptoms?
 - d. What are the differentiating features of TM type I and TM type II?
2. Differential diagnosis
 - a. What other conditions are on the list of differential diagnoses?
 - b. How can TM be differentiated from other similar retinal lesions?
 - c. What testing should be done at initial presentation before TM is diagnosed?
3. Patient management and role of the optometrist
 - a. What is the typical prognosis of TM?
 - b. How would you manage TM following the initial diagnosis?
 - c. How might your management differ with an atypical presentation?

Assessment of learning objectives

This case may be used in either the classroom or clinical setting. For classroom learning and assessment:

- The case and images can be used to enhance an advanced ocular disease course lecture.
- Live quiz features (TurningPoint, Turning Technologies LLC) or Zoom Polling (Zoom Polling, Zoom Video Communications, Inc.) during presentation may assess understanding of key concepts in real time.

- Knowledge can also be assessed through traditional examination techniques such as multiple-choice or fill-in-the-blank questions based on case presentation or pathology imaging.

For learning and assessment in the clinical setting:

- The case can be presented and discussed in small groups.
- Discussion about the use of OCT can increase the knowledge base of retinal imaging and diagnosis.
- The extensive medical history and medication list involved in this case can be used to review systemic disease and general pharmacology. One activity could include developing a table or list of this patient's systemic conditions and the corresponding medication used to treat that condition.

The recommended approach for this case would be to first review the learning objectives and read the discussion questions. Then, read the case presentation. Finally, use the Educator's Guide to answer the discussion questions and fulfill the learning objectives. This case information could also be used to generate a slide show about this condition and/or case.

Discussion

Epidemiology and pathophysiology

Torpedo maculopathy is a rare, congenital condition that predominantly presents unilaterally. It has a characteristic fundusoscopic appearance. The lesion was first alluded to in 1989 by Gass and further described in 1992 by Roseman and Gass as a hypopigmented nevus of the RPE.^{3,4} Shortly thereafter, it was renamed torpedo maculopathy.^{5,6} The lesion is occasionally referred to as paramacular coloboma, solitary hypopigmented nevus and amelanotic nevus.^{5,7,8} In 2018, Shirley et al. studied TM in the pediatric population and reported a prevalence of 2 per 100,000 in patients younger than 16.⁹ It is thought that this may be an underestimation of the true prevalence due to the asymptomatic nature of the condition. In 2021, a large systematic review found 110 case reports of TM in the literature.¹⁰

While TM is congenital, the etiology of the lesion is somewhat debated. One theory suggests that the lesion is formed due to a disruption in choroidal vasculature development.⁵ In 2010, Shields et al. outlined the most widely accepted explanation of the etiology, suggesting that the lesion is derived from the "site of the fetal bulge."¹¹ This theory postulates that a bulge forms in the temporal-macular retina during 4-6 months gestation, and while this bulge retracts during later months of gestation, it is thought that a mild residual depression remains.¹¹ It is believed that a disruption in the RPE cells occurs at this time within the residual depression, resulting in the torpedo-shaped lesion. This RPE disruption can also lead to significant attenuation of RPE cells and outer retinal layers, or a complete loss of the RPE cells altogether. Occasionally, the torpedo lesion may have an associated satellite lesion presumed to arise from the same process.⁸ Once the TM lesion is formed, it tends to be a stable, non-progressive finding. However, a few isolated case reports have outlined atypical comorbidities or progression of the lesion.^{1,2,5}

Clinical presentation and prognosis

The TM lesion is flat, oval shaped, and has well-defined borders. The vast majority of cases have presented unilaterally.¹⁰ When the fetal bulge retracts during gestation and leaves RPE disruption in its wake, the classic torpedo shape remains along with hypopigmented tissue nasally and hyperpigmented RPE hyperplasia temporally.^{4,7,11} The lesion is always temporal to the fovea due to the corresponding site of the fetal bulge. It is ellipsoid and longer horizontally than vertically, with a point facing toward the fovea matching the shape of the bulge. This classic appearance and location is pathognomonic for TM.

Fortunately, individuals with TM are typically asymptomatic.^{4,5,12} Therefore, the condition is most often

diagnosed incidentally during a comprehensive eye exam.⁷ For this reason, optometrists play a key role in the initial diagnosis and management of patients with TM. Patients typically obtain 20/20 visual acuity in the involved eye because the lesion rarely involves the fovea. There have been a few case reports in which vision is slightly reduced, but this seems to be more common with other associated pathology.⁸ Scotomas of varying depth may be present but are often not bothersome and unknown to the patient due to the congenital nature of the lesion.^{12,13}

It is well-documented that the majority of TM lesions are benign and remain stable. Few case reports have noted progression or associated complications.^{1,2,12} To the author's knowledge at this time, there has been one case report of a neurosensory retinal detachment, and three case reports of choroidal neovascular membrane (CNVM) associated with TM.^{1,2,9,15} As additional case reports emerge, there is more to be learned about potential comorbidities of this condition. For most cases, the prognosis for TM is good, and risk of vision loss or visual complication is low.⁷ Annual observation is typically recommended. Biannual or more frequent observation with home Amsler grid monitoring is recommended for atypical presentations of TM.^{1,9,12-14} Given case reports of accompanying pathology, patients should be educated on these rare, potential associations with their condition.⁹

Auxiliary testing and differential diagnosis

In addition to the funduscopic evaluation, auxiliary testing can be used to help differentiate and confirm the TM diagnosis and further classify the lesion. In particular, OCT can significantly assist in this endeavor and is key to torpedo lesion classification. TM can be classified as type I or type II based on the presence or absence of retinal and choroidal excavation on OCT; both types have outer retinal layer attenuation.¹² Type I TM is characterized by outer retinal attenuation without retinal and/or choroidal excavation, and type II TM is characterized by outer retinal attenuation with retinal and/or choroidal excavation.¹² In addition to TM classification, OCT can also help differentiate the lesion from other retinal pathology. For example, in retinal scars from toxoplasmosis, the inner and outer retina is attenuated, but in TM the inner retina will remain unchanged; this can be readily observed on OCT cross-sectional scans (**Table 1**).^{12,17} OCT angiography has shown hypopigmentation at the temporal edge of the lesion, typically in the corresponding location of the choroidal excavation and RPE hypertrophy, while hyperpigmentation is observed closer to the fovea.^{12,13} Fundus fluorescein angiography has been performed on patients with TM and has shown no leakage in areas of retinal and choroidal excavation unless an associated CNVM is present.¹⁶



Table 1. [Click to enlarge](#)

Fundus photography is a helpful tool for baseline documentation of TM and can be used to show the lesion to the patient as part of patient education. Due to the varying degree of RPE loss and retinal attenuation, visual field defects are variable in type I TM. Visual field defects are always present in type II TM due to the loss of RPE cells accompanying the choroidal excavation.¹² Baseline visual field testing is useful as it can be used to distinguish long-standing scotomas from new pathology. This is arguably more important in type I TM where the visual field defect is variable and thus may be due to a different etiology.¹² Both 10-2 and 24-2 visual field tests have been used to map TM lesions and there does not yet appear to be a clear recommendation for one test vs. the other.^{10,12}

Other lesions that may appear similar to TM include toxoplasmosis scars, congenital hypertrophy of the retinal pigment epithelium (CHRPE), histoplasmosis scars, traumatic scars and amelanotic nevi. These lesions can be differentiated based upon their appearance, location, laterality, associated visual field defects and OCT features (Table 1).^{6,17-21} A thorough case history is another important tool for differentiation. It may be helpful to ask the patient if they have had a history of toxoplasmosis infection,

blunt ocular trauma, or a history of living in the Ohio-Mississippi River Valley region due to the higher risk of histoplasmosis exposure in that geographic area.

Differentiation of similar retinal lesions:

- Toxoplasmosis scars present in the retina after a toxoplasmosis infection has subsided. These scars are full-thickness chorioretinal scars that typically present within the posterior pole and may involve the macula. Toxoplasmosis scars can be differentiated from TM due to their full-thickness chorioretinal atrophy and their non-uniform location, shape and size.¹⁷ A known history of toxoplasmosis infection can be helpful in determining the cause of retinal findings.
- CHRPE are pigmented lesions with well-defined borders. They may have a pattern of hyperpigmentation and hypopigmentation depending on the presence and location of lacunae. CHRPE lesions are most often found in the periphery of the retina and rarely present near the macula, which is a key differentiating factor from TM.^{4,18}
- Histoplasmosis is a fungal infection endemic to the Ohio-Mississippi River Valley. Patients with this condition would most likely have lived in or visited that region. Histoplasmosis scars are found in the retina after a histoplasmosis infection has resolved. These scars are often numerous and occur along with peripapillary atrophy and CNVM.¹⁹ The numerous lesions along with the two other classic findings of histoplasmosis can be used to differentiate this condition from TM.
- Chorioretinal scars may present following blunt ocular trauma. These scars have varying degrees of hyperpigmentation and hypopigmentation. They most often occur concentric to the optic nerve head and are typically vertically elongated in the pattern of a choroidal rupture.²⁰ Patients with chorioretinal scars would have a history of blunt ocular trauma to the involved eye.
- Amelanotic nevi are round non-pigmented or hypopigmented retinal lesions most often found in the retinal mid-periphery or periphery. These lesions are rarely found near the macula, thus a key differentiator from TM.²¹ Although amelanotic nevi have a variable OCT presentation, they typically lack the outer retinal attenuation and excavation seen in TM.

Treatment and management

Management of TM is typically observational and includes an annual dilated fundus exam.^{5,12} Baseline testing should be performed at initial presentation and should include fundus photography, visual field testing and OCT.^{8,12,18} OCT is particularly useful not only to determine the type of TM but also to differentiate the lesion from other similar retinal pathology and confirm the diagnosis (Table 1).¹⁶⁻²¹ Many case study authors have described TM as non-progressive, but Wong et al. postulated that the lesions may progress very slowly over many years.^{11,12} However, no treatment or intervention has been deemed necessary for retinal and/or choroidal excavation as seen in type II lesions without other associated pathology.¹² OCT can be used to confirm stability by measuring the area of outer retinal atrophy or choroidal excavation.¹² Isolated case reports of accompanying CNVM have shown good outcomes with anti-VEGF therapy.^{1,2,9} Atypical presentations of the condition may require more frequent follow-up or at-home monitoring with an Amsler grid.¹³ The presence or suspected presence of CNVM or retinal detachment necessitates referral to a retina specialist for further evaluation and management.^{1,13-15}

Patients with TM should be educated on the congenital, typically non-progressive nature of the lesion. Thorough patient education is important for rare conditions such as this to improve individuals' awareness of their diagnosis and enable better communication and continuity of care at future eye health visits. Patients should be advised of the potential rare accompanying pathology and advised to return if symptoms arise.^{1,9} Because pediatric patients are less likely to report symptoms, the importance of maintaining their routine eye exams should be emphasized. Proper identification and diagnosis of TM can be challenging due to the rarity of the condition. An increasing number of case reports, systematic reviews, and optometric education may assist with awareness and thus proper diagnosis and management of this condition.

Conclusions

Torpedo maculopathy is a rare, congenital retinal lesion with a unique fundusoscopic appearance. Patients with TM are typically asymptomatic and have 20/20 visual acuity. TM can be diagnosed upon fundusoscopic examination and can be further classified into type I or type II TM based on OCT appearance. TM is classically non-progressive and can be managed with annual observation. Few case reports have documented TM with accompanying pathology, but patients should be educated about the potential for these rare associations. It is important to differentiate TM from other progressive retinal pathology in order to determine appropriate management plans. Additionally, thorough patient education is imperative for rare conditions such as this so the patients can better communicate their diagnosis in subsequent encounters. This case outlined a typical presentation and appearance of TM type II that was managed with annual observation.

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