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Case Report

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Insights into a rare congenital abnormality: clinical and imaging features of two hiperpigmented torpedo maculopathy cases

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ABSTRACT

Torpedo maculopathy (TM) is a rare congenital, nonprogressive macular anomaly of the retina pigment epithelium (RPE) or choroid. A characteristic feature is its presentation as an isolated, asymptomatic, hypopigmented nevus in the temporal macula, often described as torpedo-shaped. Contrary to what is known, two cases of TM, both hyperpigmented, one located in the fovea, the other located in the temporal macula, and the first case accompanied by a previously unreported posterior subcapsular cataract at a young age, are presented here. These cases, along with others documented in the literature, suggest that TM can present with atypical characteristics and locations. Recognition of these lesions is important in distinguishing them from more malignant diseases such as malignant melanoma, choroidal nevus, congenital hypertrophy of RPE, and congenital toxoplasmosis.

Keywords: Congenital abnormality, maculopathy, multimodal imaging, torpedo maculopathy

INTRODUCTION

Torpedo maculopathy (TM) is a rare congenital, nonprogressive macular anomaly of the retina pigment epithelium (RPE) or choroid.[1] A characteristic feature is its presentation as an isolated, asymptomatic, hypopigmented nevus, horizontally located, in the temporal macula, often described as torpedo-shaped. TM has recently expanded with the addition of atypically located and shaped torpedo lesions to the literature and has been named "torpedo retinopathy" or even "expanded spectrum torpedo retinopathy".[2,3] With the increasing use of multimodal imaging techniques-particularly spectral domain optical coherence tomography (SD-OCT) and fundus autofluorescence (FAF) diagnosis and monitoring of torpedo maculopathy have become more precise. Typical TM is hypopigmented, and a few hyperpigmented TMs have been reported in the literature.[4,5] In this study, using multimodal imaging methods, two hyperpigmented cases and one of them with cataract at an early age, which has not been reported before, will be presented.

CASE PRESENTATION

Case 1

A 26-year-old female patient presented with bilateral complaints of decreased vision over the past few years. She reported no trauma, drug use, or gastrointestinal disease. Best corrected visual acuity (BCVA) was 5/20 (-0.50, -0.25 axis 160) in the right eye, 5/20 (-0.25, -0.50 axis 170) in the left eye. IOP was normal. Anterior segment examination revealed bilateral (more in the right eye) posterior subcapsular cataracts (PSC), with clear corneas and normal anterior chambers. Fundus examination was slightly blurred, but a solitary, oval-shaped, well-defined, partially hyperpigmented tail, torpedo-shaped lesion pointed towards the fovea in the inferotemporal macula of the right eye. FAF showed a hypoautofluorescent lesion, surrounded by a hyperautofluorescent border. SD-OCT of the right eye showed a hyporeflective subretinal cleft and defect in the RPE and ellipsoid zone corresponding to the lesion area (Figure 1). The left eye appeared normal. The patient was diagnosed with TM and was followed up for both cataract and TM.

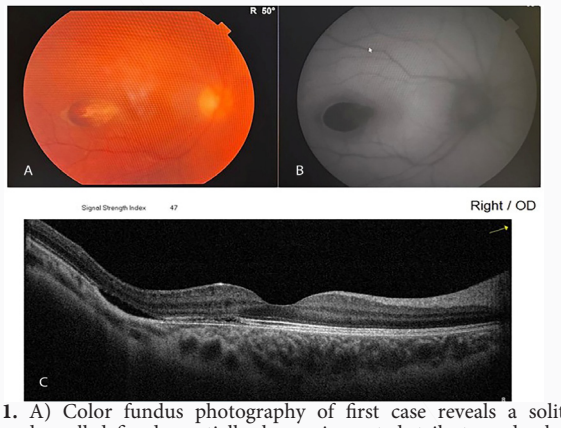

Figure 1. A) Color fundus photography of first case reveals a solitary, oval-shaped, well-defined, partially hyperpigmented tail, torpedo-shaped lesion pointing towards the fovea in the inferotemporal macula of the right eye. B) FAF demonstrates a hypoautofluorescent lesion, surrounded by a hyperautofluorescent border. C) SD-OCT of the right eye showed a hyporeflective subretinal cleft and defect in the RPE and ellipsoid zone corresponding to the lesion area

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Case 2

A 32-year-old male patient presented with a complaint of preexisting vision loss in his left eye. He had no history of trauma or systemic disease. BCVA was 20/20 (+0.25, -0.25 axis 165) in the right eye and 10/20 (-0.50 axis 160) in the left. Intraocular pressure (IOP) was normal, and anterior segment examination was unremarkable in both eyes. Fundoscopic examination revealed a torpedo-shaped lesion at the center of the fovea in the left eye, characterized by a hypopigmented peripheral zone and a hyperpigmented center. FAF demonstrated a hypoautofluorescent lesion, surrounded by a hyperautofluorescent ring. SD-OCT of the left eye demonstrated thickening of the ellipsoid zone and the presence of a cleft between the ellipsoid zone and the thinned RPE and inner choroidal excavation (**Figure 2**). Fluorescein angiography (FA) shows hyperfluorescence nasal to the lesion due to a window defect. The patient was diagnosed with torpedo maculopathy and scheduled for follow-up. We obtained an informed consent form from two patients for the procedure.

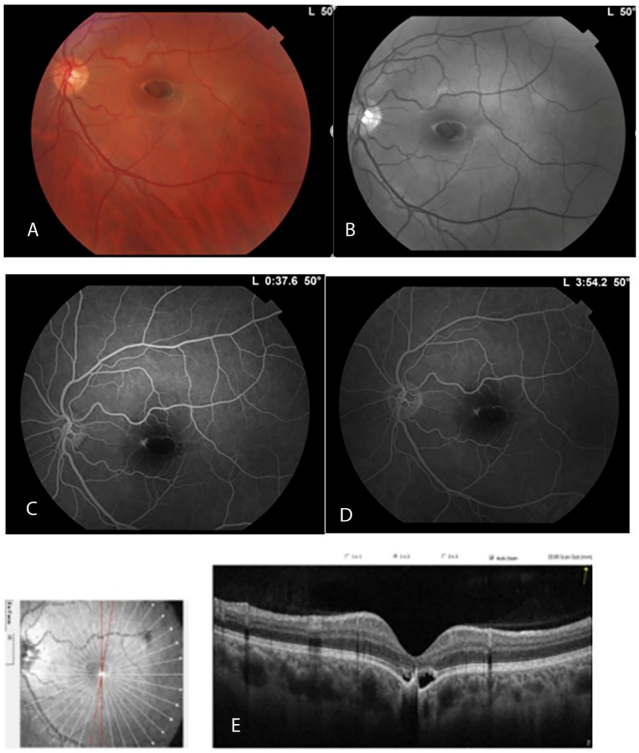

Figure 2. A) Color fundus photography of second case reveals a torpedo-shaped lesion at the center of the fovea in the left eye, characterized by a hypopigmented peripheral zone and a hyperpigmented center. B) FAF demonstrates a hypoautofluorescent lesion, surrounded by a hyperautofluorescent ring. C, D) FA shows hyperfluorescence nasal to the lesion due to a window defect. E) SD-OCT of the left eye demonstrated thickening of the ellipsoid zone and the presence of a cleft between the ellipsoid zone and the thinned RPE and inner choroidal excavation

DISCUSSION

TM is an uncommon macular anomaly characterized by a distinct, torpedo-shaped lesion within the RPE of the temporal macula. Its unique form features a rounded edge towards the temple and a pointed end directed towards the macula. It is typically asymptomatic, with an estimated occurrence of approximately 2 in every 100,000 people.[6] The exact cause of TM isn't fully understood. However, researchers suggest it's a congenital anomaly and they believe it stems from a persistent flaw in the development of the RPE within the fetal temporal bulge.[7]

The classic presentation of this lesion is a single, hypopigmented spot located in the temporal macula. Contrary to what is known, two cases of TM, both hyperpigmented, are presented here. One case is located in the fovea, and the other is located in the temporal macula. A previously unreported PSC is associated with the first case at a young age.

A few cases with hyperpigmented features have been published in the literature. Our first case has partially hyperpigmented tail in the temporal side of lesion like Yuan et al's[8] case whose explained choroidal structure characteristics. Another case whose hyperpigmentation pattern was similar to this case did not change its character and showed no progression during the 10-year follow-up.[9] In our second case, the macular lesion has a diffuse hyperpigmented center and a hypopigmented halo around it. These features are alike to the cases of Ranjith et al.[4] and Rohl et al.,[5] but while these lesions were located in the temporal macula, whereas our case was located right in the fovea. Wong et al.[10] classified OCT findings of TM into two categories: Type 1, characterized by outer retinal disturbance, and type 2, distinguished by outer retinal cavitation. In 2018, Tripathy et al.[11] defined type 3 TM by adding "inner retinal excavation" on OCT in addition to these classical TM findings. In terms of OCT characteristics, our first case exhibited type 1 TM features, while our second case displayed type 2 TM features.

The prevalence of cataracts increases with age. In addition to age, several risk factors contribute to the development of posterior PSC. These include atopy, diabetes mellitus, glaucoma, hereditary disorders, hypoparathyroidism, ocular inflammation, trauma, myopia, obesity, retinal dystrophies, solar ultraviolet (UV) radiation, steroids, and vitrectomy.[12] Our case was 26 years old and had an increasing loss of vision in the last few years. We do not know whether PSC is present at birth or whether it is added to the disease later but she did not have any of the other risk factors we mentioned. To our knowledge, this is the first case in the literature to report the coexistence of TM and PSC with a young patient.

In fact, although TM is very rarely associated with choroidal neovascularization, they are generally stable and non-progressive lesions.[9] It is important to recognize TM and to differentiate it from more aggressive lesions such as malignant melanoma, choroidal nevus, congenital simple hamartoma of the retina, congenital hypertrophy of RPE, congenital Zika virus infection and congenital toxoplasmosis. Therefore, presenting TM cases with different characteristics, as in our cases, may contribute to easier diagnosis and differential diagnosis of these cases.

CONCLUSION

In this case report, contrary to popular belief, two hyperpigmented TM cases are presented with the help of multimodal imaging methods. In one of these cases, there was PSC association, which has not been previously reported in the literature. Recognizing atypical TM cases like ours is important in distinguishing them from similar lesions with more malignant characteristics.

ETHICAL DECLARATIONS

Informed Consent

All patients signed and free and informed consent form.

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Financial Disclosure

The authors declared that this study has received no financial support.

Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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