ORIGINAL ARTICLE

Torpedo Maculopathy; Clinical and Imaging Findings

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ABSTRACT

Purpose: To report a case series of four patients with torpedo maculopathy and the review of their varied presentations with the analyses of retinal imaging.

Case series: Complete ophthalmological examination including optical coherence tomography (OCT), fluorescein angiography (FA) in one case and fundus autofluoresecence (FAF) were evaluated.

Results: All patients had ovoid, well-defined lesion located temporal to the fovea that was noticed incidentally during routine eye examination. OCT revealed attenuation of retinal layers to varying degrees and subretinal cavitation in some cases. Fluorescein angiography and FAF showed different pattern of fluorescence.

Conclusion: In this article, torpedo maculopathy cases with different clinical and imaging findings were presented.

Keywords: Torpedo maculopathy, Retina, optical coherence tomography, Fundus autofluorescence.

INTRODUCTION

Torpedo maculopathy is a benign, sharply circumscribed, ovoid lesion located at the level of the retina pigment epithelium (RPE) which typically occurs in the temporal part of the macula.[1] It has been previously named as hypomelanotic or albinotic nevus, solitary amelanotic spot and congenital hypomelanotic freckle.[1] The lesion characteristically flat, solitary, torpedo-shaped and nasal tip of the lesion which points towards the fovea. It is longer horizontally than vertically.[1] In general, ıt is asymptomatic and diagnosed accidentally on routine ophthalmic examination. Most lesions are hypopigmented, but it may contain varying degrees of pigmentation. Geographic regions of hyperpigmentation within the central hypopigmented area are also possible.[2] Torpedo maculopathy can be distinguished from other disorders of RPE, including congenital hypertrophy of RPE (CHRPE), pigmented lesions associated with Gardner syndrome, grouped pigmentation, choroidal melanoma or nevus. Estimated prevalence of torpedo maculopathy is as 2 per 100,000 population, but suggested that this is an underestimate due to lack of symptoms.[2]

In this study, we further report the varied presentation of Torpedo maculopathy in four eyes of four patients and imaging characteristics of the disease.

CASE PRESENTATION

Case 1

A 36-year-old female patient was referred to our clinic because of a lesion found in the retina during the routine ocular examination. At her first examination, visual acuity with refractive correction was 20/20 with Snellen chart in both eyes. (Table 1) There was no remarkable medical history of any systemic and ocular disease. On fundus examination, the left eye was normal and a flat, fusiform lesion with sharply defined margins and a tip pointing toward the fovea were observed in the right eye. Hyperpigmentation in the central of the lesion was observed. (Figure 1A)

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Table 1: *Patient demographics and clinical presentation.*

Patient	Gender	Age at presentation	VA	Eye	Temporal margin	Nasal margin	Pigmentation	Type on OCT	Location
Case 1	Female	36 y.o	20/20	Right	Frayed tail	Sharp point	Central hyperpigmented	Type 2	Temporal
Case 2	Female	8 y.o	20/20	Right	Frayed tail	Sharp point	Nonpigmented	Type 1	Temporal
Case 3	Female	8 y.o	20/20	Left	Round	Sharp point	Nonpigmented	Type 1	Infero-temporal
Case 4	Male	9 y.o	20/28	Left	Round	Sharp point	Nonpigmented	Type 2	Infero-temporal

Spectral-domain optical coherence tomography (OCT) revealed irregularity of the retinal layers in the correspondence of the lesion. Ellipsoid zone and external limiting membrane were distrupted and retinal pigment epithelium cleft were observed with the increased hyperreflectivity into underlying choroid. (Figure 1B)

The late phase of FA (fluorescein angiography) showed hypofluorescence in the hyperpigmented part of the lesion and hyperfluorescence in the remaining. (Figure 1C) Fundus autofluorescence (FAF) imaging revealed hypoautofluorescence corresponding to the pigmented area and isoautofluorescence with the internal focal areas of hyperautofluorescent. (Figure 1D)

Case 2

During routine eye examination, a 8-year-old girl was discovered to have a retinal lesion in the right eye. Her best corrected visual acuity (BCVA) was 20/20 in both eyes and anterior segment was normal. There were no ocular, medical history or family history of eye disease. Dilated fundus examination revealed ovoid non-pigmented, flat chorioretinal lesion in the right eye, located temporal to the fovea with the pointed tip directed towards to the fovea. The lesion was located along the horizontal raphe.(Figure 2A) Fundus examination of the left eye was unremarkable. Optical coherence tomography showed normal inner retinal layers and a localized elevation of the external

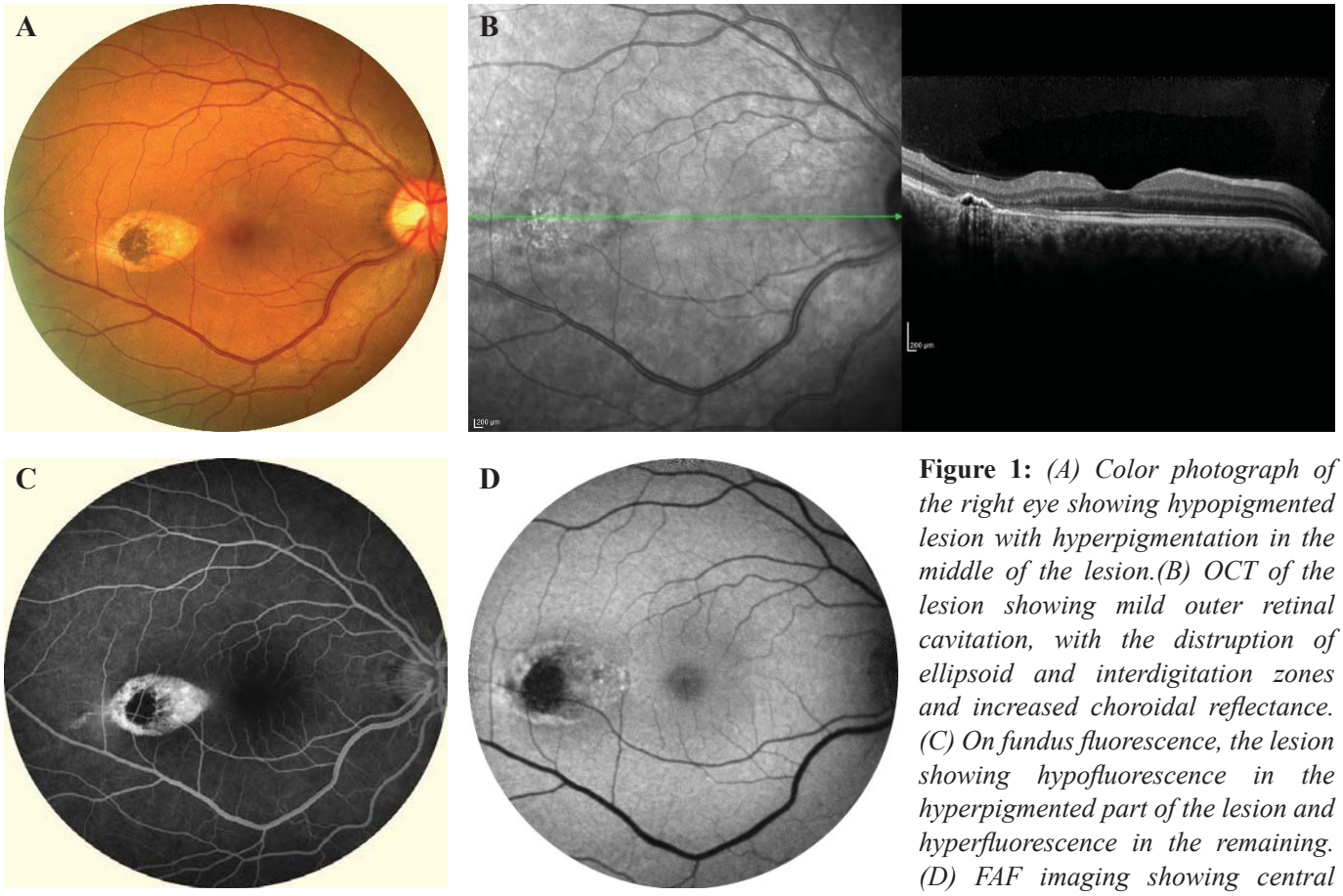

Figure 1: *(A) Color photograph of the right eye showing hypopigmented lesion with hyperpigmentation in the middle of the lesion.(B) OCT of the lesion showing mild outer retinal cavitation, with the distruption of ellipsoid and interdigitation zones and increased choroidal reflectance. (C) On fundus fluorescence, the lesion showing hypofluorescence in the hyperpigmented part of the lesion and hyperfluorescence in the remaining. (D) FAF imaging showing central hypoautofluorescence with the focal hyperfluorescent spots.*

limiting membrane (ELM) and ellipsoid zone. (Figure 2B) Fundus autofluorescence imaging demonstrated hypoautofluorescence with hyperfluorescent border in the area of torpedo maculopathy. (Figure 2C)

Case 3

A 8-year-old female child presented to our clinic for routine ophthalmic examination. Her BCVA was 20/20 in both eyes and biomicroscopy of the anterior segment was normal. She had no remarkable medical history of any ocular or systemic disease. Dilated fundus examination with a + 90-diopter lens in the left eye revealed a non-pigmented, sharply demarcated torpedo-shaped lesion, located infero-temporal macular region with the tip pointing towards the fovea. (Figure 3A) Attenuation of outer and inner retinal structures, loss of photoreceptor and RPE with choroidal hypereflectivity were observed on OCT scans and FAF imaging showed the hypoautofluorescent lesion surrounded by halo of hyperautofluorescent spots. (Figure B-C).

Case 4

A 9- year-old boy was referred to our institute due to retinal lesions found incidentally in the left eye for further examination. The fellow eye was normal. The corrected visual acuity was 20/20 in right eye and 20/28 in the left eye. Ocular and medical history was unremarkable. On dilated fundus examination, there was a hyperpigmented rounded lesion in the fovea and a flat torpedo-like lesion located infero-temporal to the fovea. (Figure 4A) A hyperreflective foveal lesion with shadowing effect, extending from the level of RPE to internal limiting membrane and a parafoveal cyst were documented on OCT scans. (Figure 4B) A large subretinal cleft was observed in correspondence of the torpedo lesion with irregularity of inner-outer segment and RPE. Inner retina and choroid were normal. (Figure 4C) On FAF, torpedo and foveal lesion were observed as hypoautofluorescence surrounded by hyperautofluorescent border. (Figure 4D)

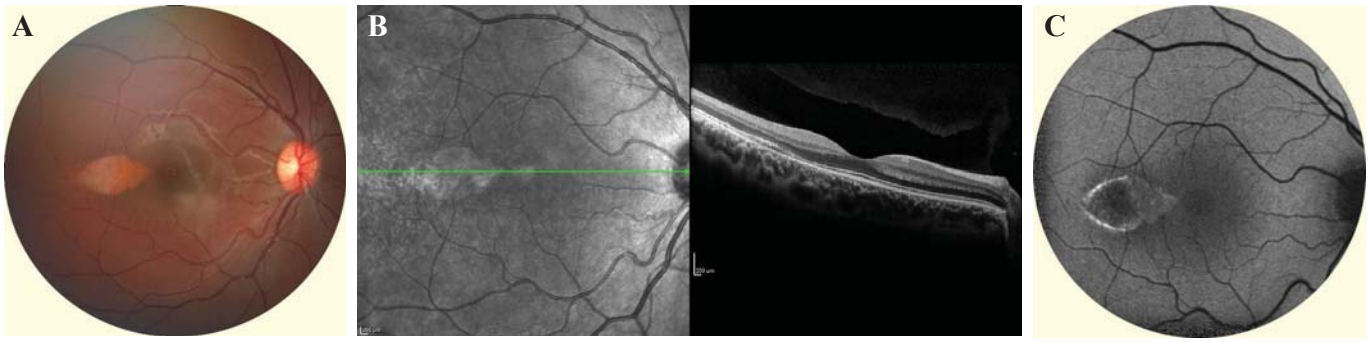

Figure 2: *(A) Fundus photograph showing hypopigmented torpedo lesion located temporal to the fovea in the right eye. (B) OCT showing a localized elevation of the ELM and ellipsoid zone with no alterations in the choroid. (C) FAF showing hypoautofluorescence lesion with a rim of hyperautofluorescence.*

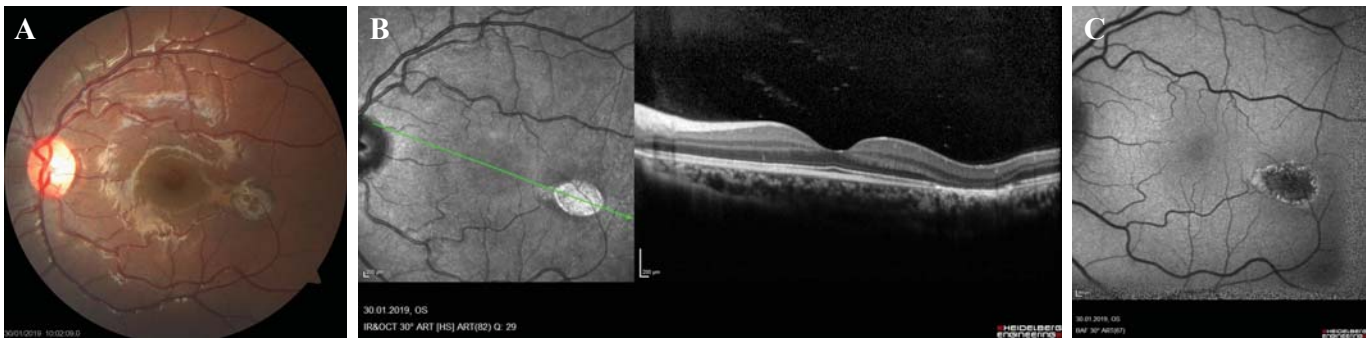

Figure 3: *(A) Fundus photo of the left eye showing hypopigmented torpedo lesion located infero-temporal to the fovea. (B) OCT showing thinning of the outer and inner retinal layers, loss of photoreceptor and RPE with the increased choroidal reflectance. (C) On fundus autofluorescence, the lesion showing homogeneous loss of autoluorescence, with a hyperfluorescent halo.*

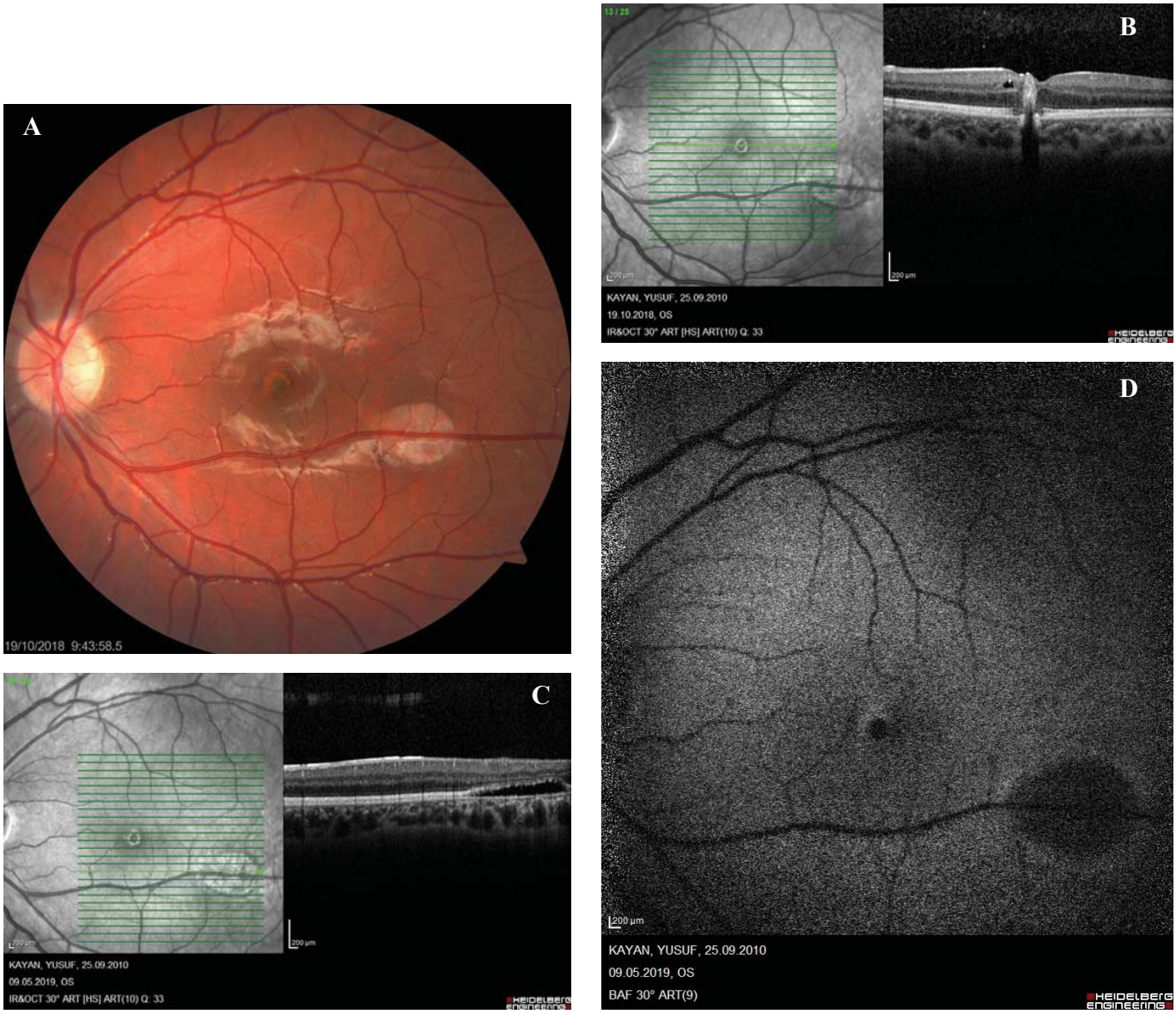

Figure 4: *(A) Fundus photograph showing a rounded lesion in the fovea and a torpedo-like lesion located infero-temporal to the fovea. (B-C) OCT revealing a hyperreflective foveal lesion with shadowing effect and a large subretinal cleft in correspondence of the torpedo lesion with irregularity of inner-outer segment and RPE. (D) On FAF, torpedo and foveal lesion were observed as hypoautofluorescence surrounded by hyperautofluorescent border.*

DISCUSSION

Torpedo maculopathy is a rare condition that is typically found during routine examination. Although ıt is almost always asymptomatic, it has been shown in some studies that there is a visual field defect in perimetry as a corresponse with the lesion.[3] It is small, flat, oval-shaped hypopigmented lesion with tip pointing to the foveola and a variably pigmented tail. Due to its characteristics shape, ıt was named as torpedo maculopathy by Daily et al.[4] On the other hand, Venkatesh et al.[5] reported two patients with torpedo lesion away from the macula and outside of the arcades. The authors proposed that torpedo retinopathy would be more appropriate terminology than torpedo maculopathy. This commonly unilateral but bilateral cases have been reported.[6,7] Although the pathogenesis of the torpedo maculopathy remains debatable, several theories have been hypothesized. Pian et al.[3] proposed that the lesion is the developmental defect in the nerve fiber layer along the horizontal raphe. Abnormal choroidal and ciliary development or persistent congenital defect of RPE in the fetal temporal bulge have also been suggested. Other hypothesis was abnormal melanin deposition in the RPE cells resulting in the hypopigmentation.[1] Another possible theory was abnormal choroidal vasculature observed

on OCT angiography (OCTA) imaging.[9,10] We did not submit OCTA evaluation of our cases as OCTA was not commercially available at the time of patient presentation.

Another controversial issue is the course of the disease. Is outer retinal cavitation inherent in the disease or developed with loss of RPE and photoreceptor over several decades? Wong et al.[11] postulated that congenital lack of melanin pigment may lead abnormal interaction RPE and outer retinal layers leading loss of ellipsoid and interdigitation zones. Continued photoreceptor degeneration leads loss of outer retinal layers and forms subretinal cavitation. The disease undergoes changes over time and is progressive. They reported retinal cavitation in older patients with an average age of 39 years; however Shirley et al.[12] reported a case series with subretinal cavitation at the age of 5 and suggested that subretinal cleft is different phenotypic presentation of the disease. We observed subretinal cleft in patients aged of 36 and 9 years old and this observation could support Shirley's theory. In addition, in our 8-year-old case (Case 3), significant photoreceptor loss was observed, but no outer retinal cavitation expected to be formed with photoreceptor loss, while in another case of the same age (Case 2), we did not observe any significant loss in the outer retinal layers and retinal cavitation. Similar to Shirley, our cases suggesting that retinal cavitation may be different phenotypic presentation of the disease. According to Wong's theory, we could deduce that the disease progressed faster in the 9-year-old case than in the 36-year-old. However the disease is typically asymptomatic and accidentally detected, our knowledge of the course and nature of the disease are limited. It is also not clear whether the disease is progressive or not.

Similar to the published reports, the lesions were located in the temporal macular region in our all cases.[1-12] The temporal aspect of the lesions may have two different configurations, including the frayed tail or the rounded margin. The frayed tail may include hypopigmentation or hyperpigmentation, while the rounded margin is more smooth and pigmented variably. In our cases, Case 1 and 2 showed hypopigmented frayed tail, whereas Case 3 and 4 had a rounded margin.

Fundus autofluorescence demonstrated a focal hypoautofluorescence corresponding to the pigmented area in Case 1, homogeneous hypoautofluorescence with hyperautofluorescent border in Case 2, 3 and 4. Different patterns of autofluorescence at torpedo lesions has also been previously reported.[9] Decreased autofluorescence is derived from atrophic RPE, while increased autofluorescence results from the build-up of lipofuscin and metabolic stress within dysfunctional RPE.[9]

Optical coherence tomography features of torpedo maculopathy are shallow outer retinal cavitation, which may appear as a neurosensory retinal detachment in some cases, and attenuation of the retinal layers to varying degrees. The subretinal cleft is thought to be formed due to photoreceptor and RPE loss.[11] The overlying retinal layers may be thinned or distrupted, but ıt can be observed normally in some cases. More recently torpedo maculopathy was classified in two types according to the pattern of abnormality in OCT.[11] The presence of outer retinal cavitation with attenuation of outer retinal structures was described to be type 2, whereas the absence of outer retinal cavitation was reported as type 1. In our series, Case 2 and 3 was categorized as a type 1 abnormality and the others were type 2. Inner retina was normal in all eyes except one. (Case 3) Thinning of outer retinal layer, interdigitation and ellipsoid zones were noted in all cases. Focal excavation of the choroid was not observed in any of our cases. Tripathy et al.[13] proposed a new classification of torpedo maculopathy and reported a type 3 lesion, which is described by excavation of inner layers, thinning of retinal layers, inner retinal hyperreflective spaces, and no subretinal cleft.

Shirley et al.[12] reported small satellite lesion temporal to the main lesion, even though torpedo maculopathy is typically described as a solitary lesion. All satellite lesion reported in the literature were small, rounded, located temporally.[12,14] In our case 4, we observed a rounded, variably pigmented lesion located in the fovea. The appearance of the lesion on FAF looked like the main lesion except it's rounded shape, but the OCT findings did not have the characteristics of the satellite lesions previously documented. There was a shadowing into the underlying choroid, not hyperreflectivity expected in torpedo lesion. Therefore, we thought that the lesion do not have an extention or association of the torpedo lesion.

Torpedo maculopathy is classically benign, but one must be alert for occasional development of choroidal neovascular membrane that could be seen in paediatric population.[12] Besides, the correct diagnosis of torpedo maculopathy is important in terms of differential diagnosis from CHRPE seen in Gardner syndrome. The RPE abnormalities associated with Gardner syndrome show a random distribution in fundus and are much smaller and more irregular in shape.[15] Some lesions have a hypopigmented halo or a hypopigmented tail that can point in any direction and lesions are usually seen bilaterally.[16] Gardner syndrome

is an autosomal dominant disease characterized by multiple colorectal polyps with at a risk of malignant transformation and by tumors outside the colon. The patients diagnosed with Gardner syndrome who have constant treat to their lives at any age and require strict follow-up.

In conclusion, torpedo maculopathy is typically asymptomatic condition with a characteristic shape and different phenotypic presentations in OCT and FAF. Further large and histopathological studies are needed to explain controversial pathogenesis and the course of the disease.

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Our presentation was approved by Ethics Committee of Numune Training Hospital, Ankara, Turkey. Neither this manuscript nor one with similar content has been published or is being considered for publication elsewhere. Also, this letter is to confirm that, the authors of this manuscript named recieved no financial support or funds and have no financial interests related to this manuscript.

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