CASE REPORT

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Torpedo Maculopathy in Newborns and First-year Outcomes

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ABSTRACT This study aimed to evaluate the clinical presentation findings and progression status of two cases with torpedo maculopathy detected during a routine newborn fundoscopic examination. The medical records of postpartum babies referred to our clinic for a routine eye examination between 2016 and 2018 were reviewed, and demographic information, clinical data, and medical history were recorded. Progression was evaluated in two cases diagnosed with torpedo maculopathy based on the fundus images taken at the first, third, and 12th months. In both cases, there was a single, flat, torpedo-like, hypopigmented lesion localized in the temporal macula at the first month. While hyperpigmentation was not detected in the lesion tail in the first month, pigmentation developed in this part in the following examinations. There was an increase in the size of the lesions up to the third month, after which the lesions no longer grew.

Keywords: Newborn fundoscopic examination; torpedo maculopathy

Torpedo maculopathy is a congenital rare anomaly of the retinal pigment epithelium (RPE), usually located in the temporal fovea, characterized by a "torpedo-shaped" appearance. It was first described by Roseman and Gass in 1992 as the asymptomatic solitary hypopigmented nevus of RPE.[1] In 1993, Daily named this lesion "torpedo maculopathy" due to its typical appearance.[2]

The characteristic fundus appearance of torpedo maculopathy is a single hypopigmented lesion with an oval shape resembling a "bullet" or "torpedo" and a wedge-shaped tail extending outward and pointing to the foveola along the horizontal raphe.[3] This characteristic appearance helps distinguish torpedo maculopathy from other conditions, such as toxoplasma scar, traumatic injury, congenital RPE hypertrophy, and Gardner's syndrome-associated RPE hypertrophy.[4]

The fundus images of our patients with torpedo maculopathy, who were diagnosed during a routine eye examination and followed up in our pediatric ophthalmology department, were recorded with the binocular indirect ophthalmoscope (HEINE OMEGA 500 LED, GERMANY) device and imaging system (DV1 digital video camera). This report aimed to present two newborn cases of torpedo maculopathy and discuss their causes and diagnostic and clinical features.

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CASE 1

The patient's parents signed informed consent. There are no conflicts of interest. The postpartum routine first-month examination of a male baby born 2,940 grams at the 38th gestational week revealed a hy-

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popigmented lesion located in the temporal fovea of the right eye with an ellipsoid border adjacent to the fovea (Figure 1). In the third-month examination, pigmentation was observed in the temporal tail part of the lesion, where the lesion was somewhat enlarged and had become more evident. At the 12th month, there was a pronounced hyperpigmentation area in the lesion but no further progression. In the dilated fundus examination, natural nervous optic nerves had a natural appearance with normal physiological pitting in both eyes. No bilateral characteristic was detected in the peripheral retinal scan. According to the family anamnesis, there was no negative medical history of the mother during her pregnancy. Serology positivity for toxoplasmosis, rubella, cytomegalovirus, and herpes simplex virus was not detected in newborn screening tests.

CASE 2

The patient's parents signed an informed consent. There is no conflicts of interest. A hypopigmented lesion with an ellipsoid border was detected in the temporal fovea of the right eye in the postpartum first-month examination of a female baby born 2,700 grams at the 38th gestational week (Figure 2). In the fundus examination, both optical discs had a natural appearance. There was no pathological finding in either eye on the peripheral retinal scan. In the third month, there was some growth in the lesion and pigmentation in the temporal tail. During the 12th month examination, torpedo maculopathy had become established with no further progression. The medical history indicated birth after a healthy pregnancy period. Serology positivity for toxoplasmosis, rubella, cytomegalovirus and her-

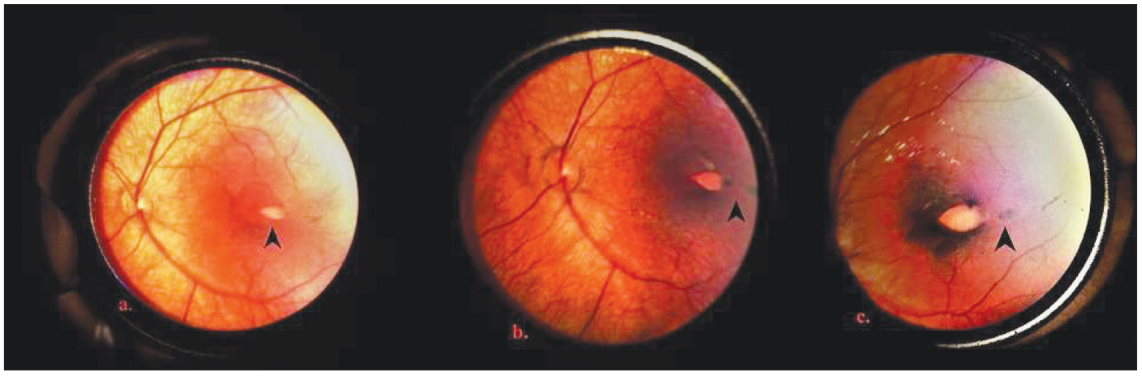

FIGURE 1: Fundus images of Case 1. a) Hypopigmented lesion at the first month, b) Pigmentation at the tail of the lesion at the third month, c) Pronounced hyperpigmentation at the 12th month.

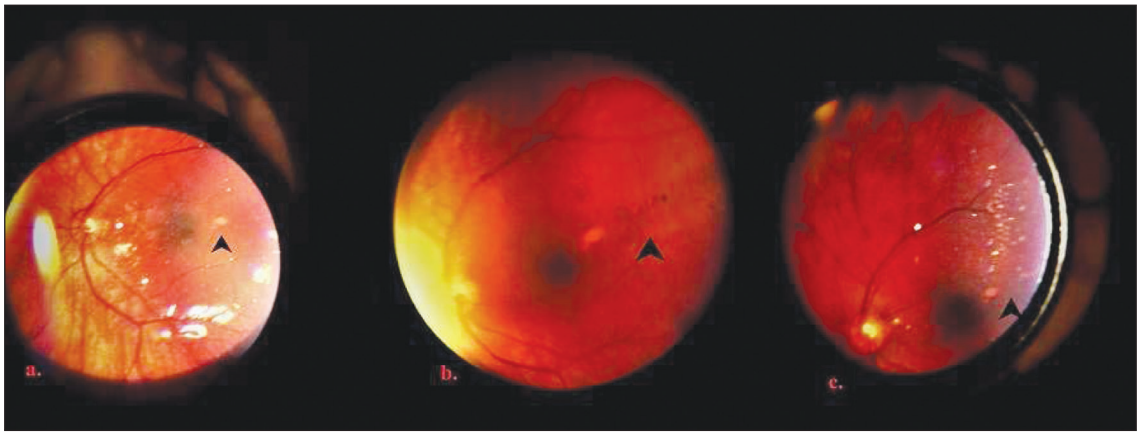

FIGURE 2: Fundus images of Case 2. a) Hypopigmented ellipsoid lesion at the first month, b) Pigmentation at the temporal tail at the third month, c) Established torpedo maculopathy at the 12th month.

pes simplex virus was not detected in newborn screening tests

DISCUSSION

Torpedo maculopathy is a rare congenital RPE disease. Although the diagnosis is made with the characteristic fundus appearance, it is essential to make a differential diagnosis to exclude other RPE diseases. The etiology of torpedo maculopathy is not yet fully known. Previous reports have suggested that a permanent defect occurs in the development of RPE as a result of abnormal nerve fiber layer, defective development of the choroid, malformation of the long posterior ciliary neurovascular bundle, or fetal temporal bulge. Although cases are generally asymptomatic, they may also present with scotoma and low vision.[4] In the differential diagnosis we made, the absence of serology positivity in the toxoplasmosis, rubella, cytomegalovirus, and herpes simplex virus panel led us to consider non-infectious causes. The lesions in our cases being single and unilateral and close to the macula helped us to exclude Gardner's syndrome, which presents with smaller and bilateral lesions further from the macula in the differential diagnosis. The lesions of our patients being unilateral and having a characteristic shape and location supported the diagnosis of torpedo maculopathy. Although lesions in torpedo retinopathy are generally unilateral, bilateral cases have also been reported in the literature, albeit rarely.[5]

In studies conducted in older age groups, it has been determined that lesions in torpedo maculopathy are generally stable, but in our two newborn cases, the lesions progressed between the first and third months and remained stable after this period. Although lesions are usually hypopigmented, cases with hyperpigmented lesions have also been described.[6] Vankatesh et al. demonstrated that the multicolor (MC) laser scanning system could be a useful tool in determining retinal and choroid layer involvement in patients with torpedo maculopathy.[7] Using MC imaging, the authors confirmed the hypothesis that torpedo maculopathy was a localized congenital anomaly of RPE pigmentation leading to outer retinal and inner choroidal degeneration. Optical coherence tomography (OCT), visual field test, and electrophysiology are methods that can support the diagnosis. In OCT, the findings of thinning of outer retinal folds below the lesion, photoreceptor loss, and sensory serous detachment can be seen in cases of torpedo maculopathy.[8]

It has been reported that in many torpedo maculopathy cases, the level of vision is not affected, but it may vary depending on the distance of the lesion to the fovea.[3] The larger lesion in our first case and its closer location to the fovea suggest that this patient was at higher risk of vision loss. Although scotoma is reported to be rare in the conventional visual field test, microperimetry shows a decrease in macular sensitivity and frequent development of central visual field defects.[3,5] In some rare cases, due to RPE and choroidal damage and neurosensory retinal detachment, follow-up is recommended annually or every six months using a dilated fundus examination, OCT, and perimetry.[9] In a five-year follow-up reported by Rohl and Vance, the presence of global hyperpigmentation, pseudo-lacuna formation, and morphological changes observed over time differed from some conventionally known definitions of torpedo maculopathy. These findings suggest that torpedo lesions may be more dynamic than previously believed and require a longer-term follow-up with multimodal imaging.[6]

Among the limitations of our study is the inability to determine the effects of lesions on the vision levels of the torpedo maculopathy cases since they were diagnosed and followed up in the neonatal period. In addition, advanced imaging methods that could help describe the lesions in detail were not used. Although our cases are the first reported in the neonatal period in the literature, gradual structural and functional changes can be better detected by examining the patients at an age when perimetry can be undertaken. We consider that our case report will contribute to the literature in terms of explaining how torpedo maculopathy lesions start and discussing the uncertainties concerning their natural course.

Source of Finance

During this study, no financial or spiritual support was received neither from any pharmaceutical company that has a direct connection with the research subject, nor from a company that pro-

vides or produces medical instruments and materials which may negatively affect the evaluation process of this study.

Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Gülay Karakuş Hacıoğlu, Alev Koçkar;* ***Design:*** *Alev Koçkar;* ***Control/Supervision:*** *Betül İlkay Sezgin;* ***Data Collection and/or Processing:*** *Gülay Karakuş Hacıoğlu, Aslıhan Doğan Dursun;* ***Analysis and/or Interpretation:*** *Alev Koçkar;* ***Literature Review:*** *Gülay Karakuş Hacıoğlu, Alev Koçkar;* ***Writing the Article:*** *Gülay Karakuş Hacıoğlu, Alev Koçkar;* ***Critical Review:*** *Betül İlkay Sezgin, Aslıhan Doğan Dursun.*

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