Torpedo maculopathy at the Site of the Fetal “Bulge”

Torpedo maculopathy was discovered in 2 children as a pointed-oval retinal pigment epithelial (RPE) defect in the temporal macula. This congenital finding could be related to the fetal temporal macular “bulge” that normally occurs at 4 to 6 months’ gestation at the same site.

There are several congenital anomalies of the RPE, including congenital hypertrophy of the RPE (CHRPE), combined hamartoma of the retina and RPE, congenital simple hamartoma of the RPE, RPE hyperplasia associated with familial adenomatous polyposis, and torpedo maculopathy. In 1992, Roseman and Gass described the features of “hypopigmented nevus of the retinal pigment epithelium,” which was later called torpedo maculopathy. This asymptomatic, torpedo-shaped defect in the RPE occurs in the temporal macular region with a pointed, torpedo-like tip directed toward the foveola. This defect closely resembles solitary CHRPE but differs in its nonrandom macular location and pointed torpedo shape. In the few reported cases, there have been no systemic associations. Herein, we describe 2 cases of torpedo maculopathy and speculate as to its embryogenesis.

Report of Cases. Case 1. On routine eye examination, a 3-year-old girl with fix-and-follow visual acuity was discovered to have a temporal macular RPE defect with a pointed-oval shape directed toward the foveola and hyperpigmented “frayed tail” appearance directed toward the ora serrata. The flat, nonpigmented lesion measured 2 mm horizontally and 1 mm vertically and was located 3.2 mm temporal to the optic disc (Figure 1). Observation was advised.

Case 2. An 11-year-old girl with uncorrected 20/20 visual acuity was discovered on routine eye examination to have a temporal macular RPE defect with a pointed-oval shape directed toward the foveola and hyperpigmented rounded temporal margin. The flat, nonpigmented lesion measured 2 mm horizontally and 1 mm vertically and was located 4 mm temporal to the optic disc (Figure 2). Toxoplasmosis titer results were negative. Observation was advised.

Comment. In 1992, Roseman and Gass described a 12-year-old boy with a small, flat, circumscribed, oval RPE lesion in the temporal macula. Additional reports confirmed the consistent pointed oval configuration and macular location of this condition (Table). Rigotti and associates reported 3 cases of asymptomatic torpedo maculopathy in a child and 2 adults. Other articles have displayed images of similar lesion dimensions measuring 2 to 3 mm horizontally and 1 mm vertically.

Congenital hypertrophy of the RPE is a flat congenital RPE lesion that appears pigmented or nonpigmented and characteristically has rounded or scalloped margins. Solitary CHRPE is located most often in the equatorial or peripheral fundus, randomly in various quadrants, and rarely in the macula (1%). Both CHRPE and torpedo macu-
Table. Published Cases of Torpedo Maculopathy

<table>
<thead>
<tr>
<th>Source</th>
<th>Sex/ Age, y</th>
<th>Eye VA</th>
<th>Location</th>
<th>Shape</th>
<th>Nasal Margin</th>
<th>Temporal Margin</th>
<th>Distance to Foveola</th>
<th>Width</th>
<th>Height</th>
<th>General Pigmentation</th>
<th>Fluorescein Angiography</th>
<th>Systemic Associations</th>
<th>Ocular Associations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Roseman and Gass, 1992</td>
<td>M/12</td>
<td>20/20</td>
<td>Temporal macula</td>
<td>Pointed oval</td>
<td>Sharp point</td>
<td>Round margin, NP</td>
<td>0.8 2 1</td>
<td>NP</td>
<td>NP</td>
<td>NP with white deep spots</td>
<td>NA</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>Teitelbaum et al, 1997</td>
<td>NA/OD</td>
<td>Temporal macula</td>
<td>Pointed oval</td>
<td>Sharp point</td>
<td>NA 1 2 1</td>
<td>NP</td>
<td>0.8 3 1.5</td>
<td>NP</td>
<td>Hyper at NP</td>
<td>Hyper at NP, hypo at P</td>
<td>NA/NA</td>
<td>NA/NA</td>
<td>None</td>
</tr>
<tr>
<td>Angioi-Duprez and Maalouf, 2000</td>
<td>NA/OA</td>
<td>Temporal macula</td>
<td>Pointed oval</td>
<td>Sharp point</td>
<td>NA 0.5 2 1</td>
<td>NP</td>
<td>0.8 3 1.5</td>
<td>NP</td>
<td>Hyper at NP</td>
<td>Hyper at NP, hypo at P</td>
<td>None/None</td>
<td>None/None</td>
<td>None</td>
</tr>
<tr>
<td>Rigotti et al, 2002</td>
<td>NA/OS</td>
<td>Temporal macula</td>
<td>Pointed oval</td>
<td>Sharp point</td>
<td>Frayed tail</td>
<td>Round margin, linear P</td>
<td>0.8 3 1.5</td>
<td>NP</td>
<td>Hyper at NP</td>
<td>Hyper at NP, hypo at P</td>
<td>NA/NA</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>Mahieu and Mathis, 2003</td>
<td>NA/OS</td>
<td>Temporal macula</td>
<td>Pointed oval</td>
<td>Sharp point</td>
<td>Tail present</td>
<td>Round margin, linear P</td>
<td>0.8 3 1.5</td>
<td>NP</td>
<td>Hyper at NP</td>
<td>Hyper at NP, hypo at P</td>
<td>NA/NA</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>Current study</td>
<td>F/3 OD</td>
<td>F+F</td>
<td>Temporal macula</td>
<td>Pointed oval</td>
<td>Frayed tail</td>
<td>NP surrounding &quot;normal&quot; RPE hyperpigmentation</td>
<td>0.8 3 1.5</td>
<td>NP</td>
<td>Hyper at NP</td>
<td>Hyper at NP, hypo at P</td>
<td>None/None</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>Current study</td>
<td>F/11 OD</td>
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<td>Temporal macula</td>
<td>Pointed oval</td>
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<td>Round margin, linear P</td>
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<td>NP</td>
<td>Hyper at NP</td>
<td>Hyper at NP, hypo at P</td>
<td>None/None</td>
<td>None</td>
<td>None</td>
</tr>
</tbody>
</table>

Abbreviations: F: fix and follow; hyper, hyperfluorescent; hypo, hypofluorescent; NA, not available or photographs do not clearly depict feature; NP, nonpigmented; P, pigmented; RPE, retinal pigment epithelium; VA, visual acuity.

* Illustrations included only 1 fundus photograph and no fluorescein angiograms. Data are based on clinical description. Exact ages not given, but indicated that there were 1 child and 2 young adults.

Torpedo maculopathy is presumed to be congenital RPE abnormalities, but its random distribution and rounded appearance is unlike torpedo maculopathy. The RPE abnormalities associated with familial adenomatous polyposis and Gardner syndrome are also similar to torpedo maculopathy, but those with familial adenomatous polyposis manifest a random distribution in the fundus and are often much smaller and more irregular in shape. In the published cases of torpedo maculopathy and our 2 current cases, there seems to be similarities in the clinical features of this condition in that all illustrations have shown a nonpigmented RPE lesion within the temporal region of the macula, ranging from immediately underneath the foveola to 1 mm from the foveola and approximately 2 to 3 mm in horizontal diameter and 1 mm in vertical diameter (Table). In all cases, the lesion was oval with a characteristic point aimed toward the foveola. There have been notable differences, however, in the temporal aspect with 2 alternative configurations that include a “frayed tail” or a rounded margin. The frayed tail was composed of either linear or dotted hyperpigmentation and hypopigmentation. The rounded margin was smooth and composed of either linear, rounded, or no hyperpigmentation at the temporal margin. In our 2 cases, one showed the frayed tail appearance, whereas the other had a rounded margin.

The etiology of torpedo maculopathy remains speculative and some have credited abnormal choroidal development or ciliary vasculature development leading to the localized, nonprogressive RPE lesion. The uniform location and size of this condition points toward a congenital defect at a precise time during fetal development of the RPE. The RPE is derived from the outer wall of the optic cup. Full pigmentation of the RPE is achieved by the 10-mm stage (fifth week). In 1969, Streiten studied fetal RPE development using 16 fetal eyes at various gestational months, 4 eyes from 3 neonates, and 35 pairs of eyes from those aged 2 months to 74 years. She noted that the fetal RPE cells increase in size from the ora serrata to the macular region. In the macular region, there was a prominent cone-shaped bulge in the temporal posterior pole of the eye approximately 4 mm from the optic disc and centered slightly temporal to the fovea. The bulge was first noted in the 4-month fetus and gradually enlarged to a staphylomatous or sclerectasia appearance by 6 months and gradually lessened to a 3.5-mm shallow concavity by 8.5 months. At term, a “slight residual depression” was found 4 mm temporal to the optic disc, again slightly temporal to the fovea. Retinal pigment epithelial cell count in the 4-month fetal macula was 45 per field, and in the temporal bulge there was an unusually high RPE count of 70 per field. In comparison, the adult macula showed an RPE cell count of approximately 30 per field. The dense RPE cellularity in the bulge apex gradually disappeared as the bulge matured. According to Streiten, the RPE cells in the bulge concavity were large and...
dividing rapidly. Streeten postulated that the temporal bulge was designed to “fully expand the macular area by the 8th month gestation.”

This prominent feature of fetal RPE development correlates in location and size with torpedo maculopathy. Based on these observations, torpedo maculopathy could represent a persistent defect in the development of the RPE in the fetal temporal bulge.

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Ophthalmological Ephemera

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