An Insight Into the Pathogenesis of Optic Disc Pit–Associated Maculopathy With Enhanced Depth Imaging

Jaitra P. Gowdar, DNB; Bindu Rajesh, MS; Anantharaman Giridhar, MS; Mahesh Gopalakrishnan, MS; Rameez Hussain, MS; Thomas Thachil, MS

OPTIC NERVE PIT is a rare congenital abnormality presenting with excavation of optic nerve head probably arising as a result of incomplete closure of the superior end of the embryonic fissure. The prevalence of optic disc pit was estimated to be 1 in 11 000 by Reisin in 1908.1 Although reported earlier, the relation between optic disc pit and maculopathy was emphasized by Petersen2 in 1958. Gass3 proposed cerebrospinal fluid as the source of fluid whereas Sugar4 suggested liquefied vitreous to be the source. Brown et al5 demonstrated the connection between vitreous cavity and macular fluid in their experimental study on collie dogs. However, this has not been demonstrated in humans.

Enhanced depth imaging (EDI) technology of spectral-domain (SD) optical coherence tomography (OCT) has been used previously to image the choroid and deeper structures such as the lamina cribrosa and postlaminar structures. In our study, we tried to assess the morphology of the deeper structures in the optic nerve head pit using the EDI technique.

Methods
A prospective observational study of consecutive patients with optic disc pit maculopathy was carried out in our institute between January 2013 and November 2013. Institutional review board approval was obtained from Giridhar Eye Institute, Kerala, India. Patient consent was waived because the hospital requires only when patients undergo invasive diagnostic procedures; this study was observational and included non-invasive fundus photography and OCT. Also, per the Ethical Guidelines for Biomedical Research on Human Participants of the Indian Council of Medical Research, the requirement for informed consent is waived if the research involves minimal risk to patients and if patients' privacy is protected.

Cystoid spaces with intervening septa in the nerve fiber layer and/or ganglion cell layer were defined as inner layer retinal schisis (ILRS) while cystoids in the outer plexiform layer and/or outer nuclear layer were defined as outer layer retinal schisis (OLRS). All enrolled patients underwent complete ophthalmic evaluation. In vivo anatomy of disc and macula was scanned using SD-OCT (Spectralis, Heidelberg Engineering). Scanning was done using 30° retinal windows with a central internal fixing light to center the macula and nasal target to center the disc. The macula was scanned in volume mode with a minimal area of 20° × 15°, density of 240 μm, each line section having 512 A-scans, and with a minimum automatic real time of 9 frames. Enhanced depth imaging mode with a minimum area of 15° × 10°, density of 60 μm, 384 A-scans, and automatic real time of 16 frames was selected to scan disc. The morphological characteristic of the retina and the prelaminar, lamina

Importance Optic disc pit with associated maculopathy is a known entity. However, controversy exists regarding the source of subretinal fluid in these cases. In our series, we attempted to analyze the morphologic changes seen in the optic disc pit and evaluate the source of subretinal fluid.

Observations In this prospective observational case series of 4 patients with optic pit maculopathy, a complete ophthalmic evaluation, with fundus color photography and enhanced depth imaging spectral-domain optical coherence tomography scanning of the optic disc, was carried out between January 2013 and November 2013. The optical coherence tomographic section was mapped with infrared image and color photography, and the characteristics of the retina and optic nerve head were analyzed. All the cases had outer layer retinal schisis; 2 of them had associated serous macular detachment while inner retinal schisis was present in 3 cases. A hyporeflective tract was observed in our study connecting the retinal schisis cavity and gap in the lamina cribrosa corresponding to the optic pit.

Conclusions and Relevance In our study, we demonstrated the connectivity between retinal schisis and the gap in the lamina cribrosa present in the optic disc pit, supporting the hypothesis of cerebrospinal fluid as the source of subretinal fluid.

Published online February 12, 2015.

Author Affiliations: Giridhar Eye Institute, Kerala, India.
Corresponding Author: Bindu Rajesh, MS, Giridhar Eye Institute, Ponneth Temple Road, Kadavanthra, Cochin 682020, Kerala, India (docbindu@gmail.com).
Table. Demographic and Clinical Features of Patients

<table>
<thead>
<tr>
<th>Patient No./Sex/Age, y</th>
<th>Eye</th>
<th>Visual Acuity</th>
<th>Pit Location</th>
<th>Maculopathy</th>
<th>Tract Connection</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/Female/teens</td>
<td>Right</td>
<td>20/80</td>
<td>Temporal</td>
<td>SMD+OLRS+ILRS</td>
<td>ILRS</td>
</tr>
<tr>
<td>2/Male/40s</td>
<td>Left</td>
<td>20/60</td>
<td>Temporal</td>
<td>SMD+OLRS</td>
<td>OLRS</td>
</tr>
<tr>
<td>3/Female/40s</td>
<td>Right</td>
<td>20/400</td>
<td>Temporal</td>
<td>ILRS+OLRS</td>
<td>ILRS</td>
</tr>
<tr>
<td>4/Male/60s</td>
<td>Right</td>
<td>20/400</td>
<td>Temporal</td>
<td>ILRS+OLRS</td>
<td>ILRS</td>
</tr>
</tbody>
</table>

Abbreviations: ILRS, inner layer retinal schisis; OLRS, outer layer retinal schisis; SMD, serous macular detachment.

All eyes revealed temporally located optic disc pits. Outer layer schisis was observed in all the eyes, with 3 eyes having additional inner layer schisis. Serous macular detachment (SMD) was seen in only 2 eyes. Enhanced depth imaging of the optic disc revealed a hyporeflective homogenous tract connecting the disc pit to the retinal schisis—either the ILRS or the OLRS in all the eyes.

Report of Cases
Case 1
A teenaged girl with blurring of vision in the right eye of 2 years' duration presented with best-corrected visual acuity (BCVA)
of 20/80 OD and 20/20 OS. Fundus examination revealed a dirty gray oval pit (Figure 1A) with a SMD. Optical coherence tomography revealed the presence of a large SMD with both ILRS and OLRS. Enhanced depth imaging through the pit revealed an abruptly ending hyperreflective lamina cribrosa. The gap in the lamina cribrosa appeared to be filled with a heterogeneous reflective lesion suggestive of glial tissue. The connectivity between the inner layer schisis and the optic disc pit is seen as a hyporeflective tract running straight down along the temporal disc margin (Figure 1B).

Case 2
A man in his 40s with BCVA of 20/20 OD and 20/120 OS revealed a temporal optic disc pit (Figure 1C) with a large SMD in the left eye. Optical coherence tomographic imaging of the macula revealed an OLRS with a SMD. Enhanced depth imaging of the optic disc revealed a deep cup and a pit with deeply placed lamina cribrosa (Figure 1D). A hyporeflective homogeneous tract is visible at the temporal aspect connecting a prelaminar cavity to the OLRS.

Case 3
A woman in her 40s with hypothyroidism and defective vision of 10 years presented with BCVA of 20/400 OD and 20/20 OS. Fundus examination of the right eye revealed a temporal optic disc pit with macular schisis (Figure 2A). Optical coherence tomography of the macula revealed both ILRS and OLRS. Enhanced depth imaging of the optic nerve head revealed a heterogeneous reflective lesion filling the pit area. Lamina cribrosa was observed to be slanting and deformed (Figure 2B), with the nasal side placed higher than the temporal side. A hyporeflective cystoid tract was observed from the inner retinal layer to a cavity within the optic disc, extending through the defect in lamina cribrosa.

Case 4
A man in his 60s with defective vision in the right eye of 3 years’ duration presented with BCVA of 20/400 OD and 20/30 OS. Optic disc pit and macular schisis visible clinically were confirmed on OCT (Figure 2C). Enhanced depth imaging revealing a heterogeneous reflective lesion filling the optic pit. A thin...
hyporeflective tract is seen connecting the ILRS and the cavity within the optic disc pit (Figure 2D).

Discussion

In 1988, Lincoff et al.6 identified the patterns of retinal schisis by slitlamp biomicroscopy in patients with optic disc pit maculopathy prior to the advent of OCT. Connection between the schisis cavity and the optic disc has been reported by many investigators.9

Prelaminar cavities were visible in our cases, with case 3 having a large one. The difference in the cavity size could probably be explained by the fact that lamina cribrosa, prelaminal tissue integrity, and resistance of the retinal layer to separation influence the formation, size, and shape of the prelaminar cavity.10 The heterogeneous reflective lesions filling the optic pit were suggestive of glial tissues or the rudimentary retinal tissues as described in histopathological studies.11

None of our cases revealed a break in retinal continuity in the inner layers in any of the OCT sections around the disc. The minimum age at presentation in our series was 13 years. Liquefied vitreous being rare in this age group further supports the theory of cerebrospinal fluid as the source of subretinal fluid.12

Histological studies on temporally and inferotemporally located optic pits have shown lamina cribrosa defects at the location of pit possibly explaining the association of optic disc pit maculopathy.13 As peripheral lamina cribrosa and peripapillary sclera separates the vitreous cavity from the subarachnoid space, any gap in this area can cause cerebrospinal fluid leak into the optic disc.12 Owing to their central location and distance from the subarachnoid space, central pits are usually unassociated with maculopathy.15

Conclusions

In our study using EDI, we demonstrated the connectivity between the schisis cavity in the retina and the gap in the lamina cribrosa present in the optic disc pit, supporting the hypothesis of cerebrospinal fluid as the source of subretinal fluid. Although the small sample size limits the results of our study, the demonstration of connecting tract on EDI provides an insight into the pathogenesis of optic disc pit maculopathy and can pave the way for future treatment modalities for closure of the tract.