PERSPECTIVE

Pathogenesis and Treatment of Maculopathy Associated With Cavitary Optic Disc Anomalies

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• PURPOSE: To propose a unifying theory regarding the pathogenesis of maculopathy associated with cavitary optic disc anomalies and to describe a rational approach to achieving a permanent cure in affected eyes.
• DESIGN: Interpretive essay.
• METHODS: Review and synthesis of selected literature, with interpretation and perspective in relating pathoanatomic features to pathogenesis and treatment.
• RESULTS: Congenital cavitary anomalies of the optic disc, including typical coloboma, optic pit (and other atypical colobomas), morning glory anomaly, and extrapapillary cavitation, are associated with an enigmatic maculopathy characterized by schisis-like thickening and serous detachment. The unifying anatomic theme of these anomalies is the presence of a scleral (or lamina cribrosa) defect permitting anomalous communications between intraocular and extraocular spaces. These communications enable the critical pathogenic mechanism responsible for the maculopathy, namely, dynamic fluctuations in the gradient between intraocular and intracranial pressures that direct the movement of fluid (vitreous humor or cerebrospinal fluid) into and under the retina. Vitreous traction does not seem to play a significant pathogenic role. Permanent cure of the maculopathy requires either elimination of the translaminar pressure gradient or closure of the pathway for fluid flow into the retina. We advocate carefully titrated juxtapapillary laser photocoagulation followed by vitrectomy with gas tamponade for creation of a permanent intraretinal and subretinal fluid barrier.
• CONCLUSIONS: The peculiar features of cavitary optic disc maculopathy can be explained only by considering the pressure gradients that develop along anomalous communications between intraocular and extraocular spaces. A permanent cure for this condition can be achieved by closing the pathway for fluid migration from the cavitary lesion into and under the retina. (Am J Ophthalmol 2014;158:423–435. © 2014 by Elsevier Inc. All rights reserved.)

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A PECULIAR AND ENIGMATIC MACULOPATHY commonly occurs in association with several congenital cavitary anomalies of the optic disc, including typical optic nerve coloboma, optic pit (and other atypical colobomas), morning glory anomaly, and extrapapillary cavitation. Although usually described as distinct clinical entities, these anomalies likely fall within a spectrum with variable pathoanatomic features but similar embryogenesis and pathologic sequelae.

The pathogenesis of serous maculopathy complicating cavitary disc anomalies is as intriguing as it is controversial, and for many decades has defied attempts to understand it clearly. This lack of clarity has spawned a diversity of treatment approaches with variable efficacy. Attempting to understand the fluid origin and pathophysiology of this condition informs us about the structure of the optic disc and juxtapapillary retina and the dynamic forces that influence these tissues. In this Perspective, we discuss the varying clinical presentations of the cavitary disc anomalies and associated maculopathy, the probable pathophysiologic mechanisms involved, and key considerations regarding management.

CLASSIFICATION OF CONGENITAL CAVITARY OPTIC DISC ANOMALIES

TYPICAL COLOBOMA OF THE OPTIC NERVE IS A CONGENITAL excavation located in the inferonasal aspect of the disc (Figure 1, Top left). It is typically a sporadic and unilateral condition. The juxtapapillary tissues inferior to the disc may be involved. Often, the affected eye is otherwise normal, and visual field loss is confined to the site of the defect. Localized serous maculopathy may develop in eyes with optic nerve coloboma,[1] whereas extensive rhegmatogenous retinal detachment more likely occurs in eyes with large associated chorioretinal coloboma.[2]

Optic disc pits are focal cavitations isolated to the disc that are typically small and temporally located, but can also be quite large and involve other portions of the disc. They lie on a spectrum of anomalies often referred to as *atypical optic nerve colobomas*.[3–5] Although classic optic pits usually are unilateral and appear as focal

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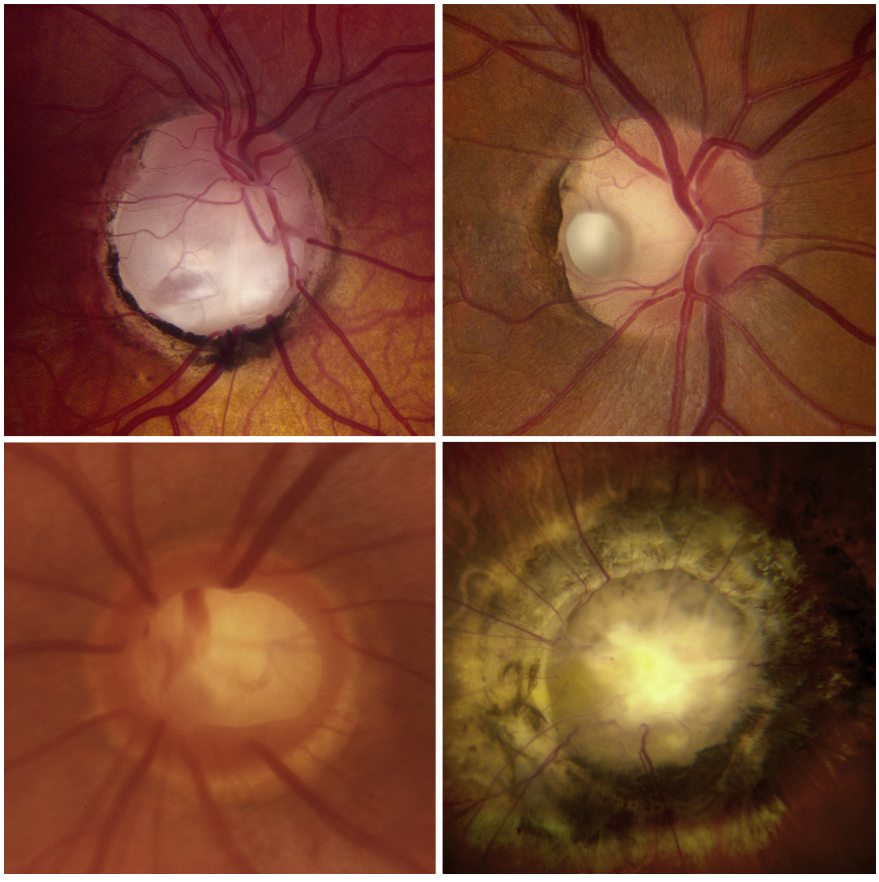

FIGURE 1. Fundus photographs showing the spectrum of cavitary disc anomalies: (Top left) typical coloboma, (Top right) classic optic pit, (Bottom left) centrally located atypical coloboma, and (Bottom right) morning glory anomaly.

grayish cavitations (Figure 1, Top right), when atypical colobomas are bilateral, large, and centrally located, they can be mistaken for glaucomatous optic cupping (Figure 1, Bottom left). Optic pits may cause congenital field defects because of associated nerve fiber layer defects. Serous maculopathy develops in more than 50% of cases, particularly in eyes with large and temporally located excavations.[6,7]

Morning glory disc anomaly is a unilateral markedly enlarged optic papilla with funnel-shaped excavation involving the optic disc and peripapillary retina (Figure 1, Bottom right). It features a characteristic radial arrangement of retinal vessels emanating from the disc, glial hyperplasia over the center of the disc, a rim of elevated peripapillary pigmented tissue, and traction on adjacent retina.[8] Affected patients typically have poor vision.

Because of its inferonasal location, typical optic disc coloboma is attributed to faulty closure of the embryonic ocular fissure. However, several lines of evidence suggest that all of the congenital cavitary disc anomalies associated with macular detachment may share similar embryogenic mechanisms. Because the normal embryonic fissure extends along the inferior aspect of the globe and completely surrounds the optic papilla at its most posterior extent, it is anatomically plausible that faulty closure of this fissure could result in the diversity of cavitary disc lesions under discussion.[9] Furthermore, histopathologic specimens demonstrate similarities between optic pit, morning glory syndrome, and typical coloboma.[8] In each case, dysplastic retina is herniated posteriorly through a defect in the lamina cribrosa, juxtapapillary sclera, or both, ranging from a focal defect in optic pits to a circumpapillary defect in morning glory anomalies. Finally, both optic pit and morning glory discs frequently occur in association with typical coloboma in the same or contralateral eye.[9] In 1 extended family spanning 5 generations with an autosomal dominant inheritance pattern, 35 members had a phenotypically diverse range of optic disc anomalies, including disc pit, coloboma, and morning glory syndrome.[4]

In addition to these anomalies involving the optic nerve itself, cavitary anomalies that lie separate from but near to the optic nerve occasionally can cause a maculopathy identical to that seen with cavitary disc lesions. On optical coherence tomography (OCT) imaging, the pathoanatomic features of such lesions, as with the disc anomalies, seem to involve herniation of abnormal retinal tissue through a scleral defect.[10]