DYNAMIC ATYPICAL OPTIC NERVE COLOBOMA ASSOCIATED WITH TRANSIENT MACULAR DETACHMENT

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ABSTRACT

Purpose: Macular schisis or detachment is frequently observed in eyes with optic pits or colobomas. Although spontaneous resolution of the maculopathy has been reported, concurrent changes in the optic nerve coloboma have not. We report three cases of atypical optic nerve colobomas in which dynamic optic nerve changes coincide with the development and subsequent resolution of the associated maculopathy.

Methods: We reviewed the records of three patients with dynamic optic nerve changes associated with maculopathy. All patients were observed for at least 6 months. Fundus photography and fluorescein angiography were used to document the optic nerve and macular changes.

Results: Three patients were noted to have macular detachments without apparent optic nerve excavation. With observation, the maculopathy spontaneously resolved in each case. We documented concurrent optic nerve changes whereby atypical optic nerve colobomas became apparent over several months in all cases. In one case, we noted the simultaneous development of maculopathy in association with obscuration of a prior disc anomaly. None of the eyes had a posterior vitreous detachment. We could not identify any associated systemic conditions or reproduce the findings with external stimulation. Initial Snellen acuity ranged from 20/60 to 20/200. Final Snellen acuity ranged from 20/20 to 20/40.

Conclusions: Fluctuating optic nerve changes may occur in the setting of atypical optic nerve coloboma and associated maculopathy. In cases of macular schisis or detachment where an optic nerve coloboma is not readily apparent, and no other causes are identified, consideration of a period of observation prior to therapeutic intervention seems appropriate.

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INTRODUCTION

Though infrequently encountered by most ophthalmologists, optic nerve pits and optic nerve colobomas (typical and atypical) are well-known cavitary optic disc anomalies.[1,2] Maculopathy characterized by macular schisis, outer layer detachments, and occasionally outer layer holes is frequently associated with cavitary optic disc anomalies.[1,3,4] The etiology of the maculopathy and source of the subretinal fluid are unknown, but both systemic and intraocular factors have been proposed.[3-9]

Fluctuations, and even spontaneous resolution, of the described maculopathy can occur but are uncommon.[1,3,5,10] Though other cavitary optic disc anomalies have rarely been reported to fluctuate in appearance,[11-13] we are unaware of any previous reports of fluctuating optic pits or atypical colobomas. We present three unique cases in which the fluctuating appearance of an atypical optic nerve coloboma is associated with spontaneous resolution of the associated maculopathy.

CASE REPORTS

Case 1

A 40-year-old white woman presented with a 1-week history of decreased vision in her right eye. Family history was negative for colobomas and optic nerve disease, but one brother had a history of cataract and retinal detachment. Best-corrected acuity was measured at 20/200 in both eyes. The visual acuity of the left eye had slowly deteriorated 4 years previously as a result of progression of low-tension glaucoma. At presentation, the patient was using timolol maleate ophthalmic solution (Timoptic) and brinzolamide ophthalmic suspension (Azopt) in both eyes. Results of previous neuroimaging with computed tomography and magnetic resonance imaging were normal. When first examined, the optic nerve of the right eye was normal in size and color and contained a small slit-like cup (Figure 1A). Neither an optic pit nor peripapillary pigment changes were seen. A large macular schisis with a large outer layer detachment was noted. The fovea appeared cystic, and no outer layer hole could be identified. A posterior vitreous detachment could not be identified. Late frames of the fluorescein angiogram demonstrated mild disc hyperfluorescence. The left disc was of normal size without peripapillary pigmentary changes and contained a large, uniformly deep cup (cup-disc ratio of 0.95). The macula appeared normal. We could identify the base of the cup, and no distinct excavation was seen. Observation was recommended.

Over the next 6 months, the acuity of the right eye improved to 20/40. The schisis and outer layer detachment resolved, but macular retinal pigment epithelial changes corresponding to the area of the schisis were still present. A distinct optic nerve cup containing a central excavation was now visible (Figure 1B).

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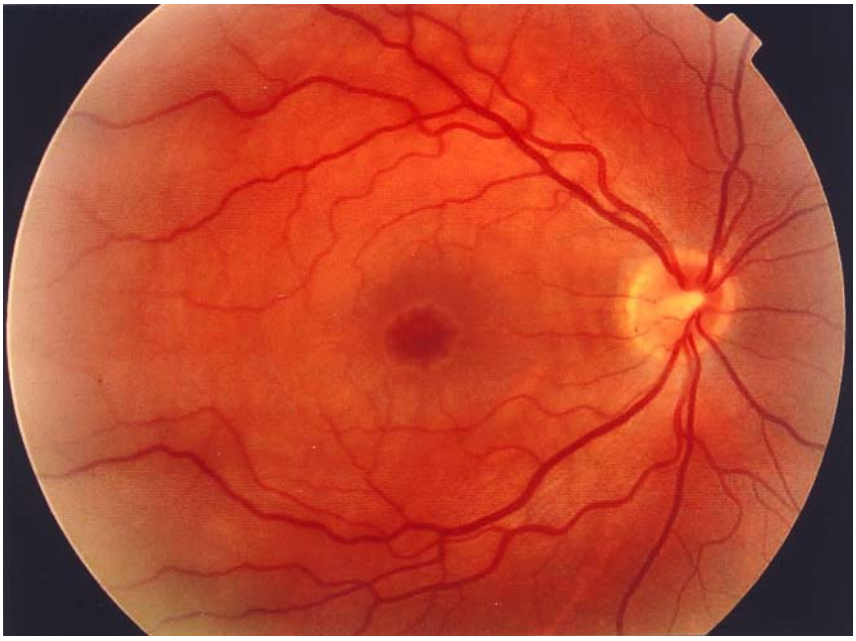

FIGURE 1A

Case 1. Dynamic atypical optic nerve coloboma with transient macular detachment in a 40-year-old woman with a 1-week history of decreased vision in the right eye. Fundus photograph of the right eye shows an apparently normal nerve with a small slit-like cup and associated macular schisis with outer layer macular detachment. No optic pit or coloboma was visualized. Visual acuity was 20/200.

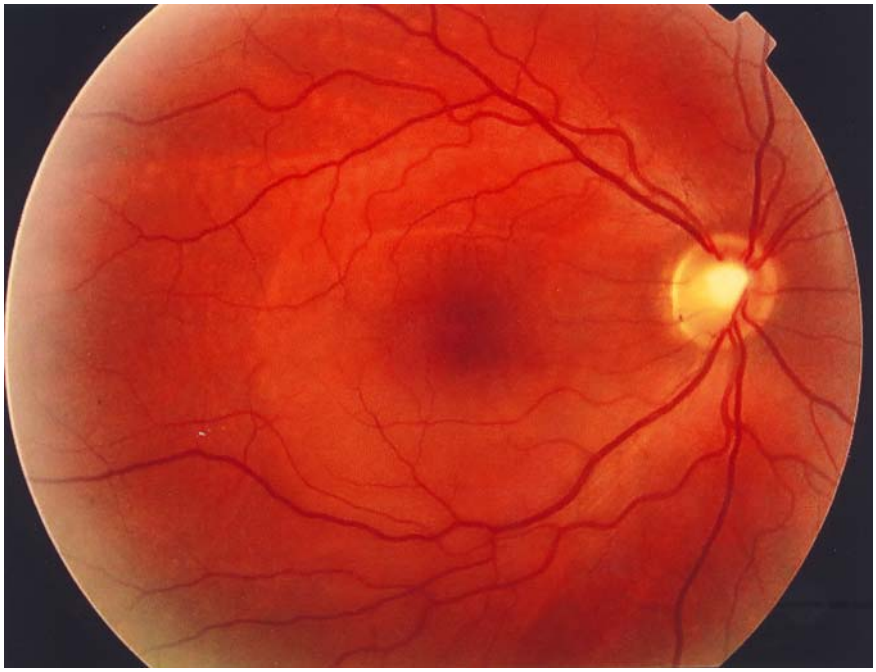

FIGURE 1B

Case 1, right eye, 6 months after photograph in Figure 1A was taken. No treatment had been administered. Optic nerve cavitation is apparent but the macular schisis and outer layer detachment have resolved. Visual acuity improved to 20/40.

Funduscopic examination of the left eye was unchanged throughout the course of observation. Ocular coherence tomography (OCT) was not available. The funduscopic appearance could not be reproduced by changes in body position or by light stimulation of the eye.

Case 2

A 30-year-old white man presented with a 1-week history of blurred vision in the left eye. Family history was negative for colobomas and optic nerve disease. Corrected acuity measured 20/20 in the right eye and 20/160 in the left eye. The patient had previously undergone bilateral myopic photorefractive keratectomy and had an uncorrected acuity of 20/20 in both eyes. The optic nerve of the right eye appeared normal with a cup-disc ratio of 0.3. The floor of the cup was clearly seen, and the macula appeared normal. The left optic nerve was normal in size without peripapillary pigment changes. The nerve was pink with slightly blurred margins and contained a thin, centrally located, slit-like cup without a visible excavation (Figure 1C). A large macular schisis cavity with a smaller, centrally located outer layer detachment was observed. A posterior vitreous detachment could not be identified. Late frames of the fluorescein angiogram revealed increasing peripapillary hyperfluorescence.

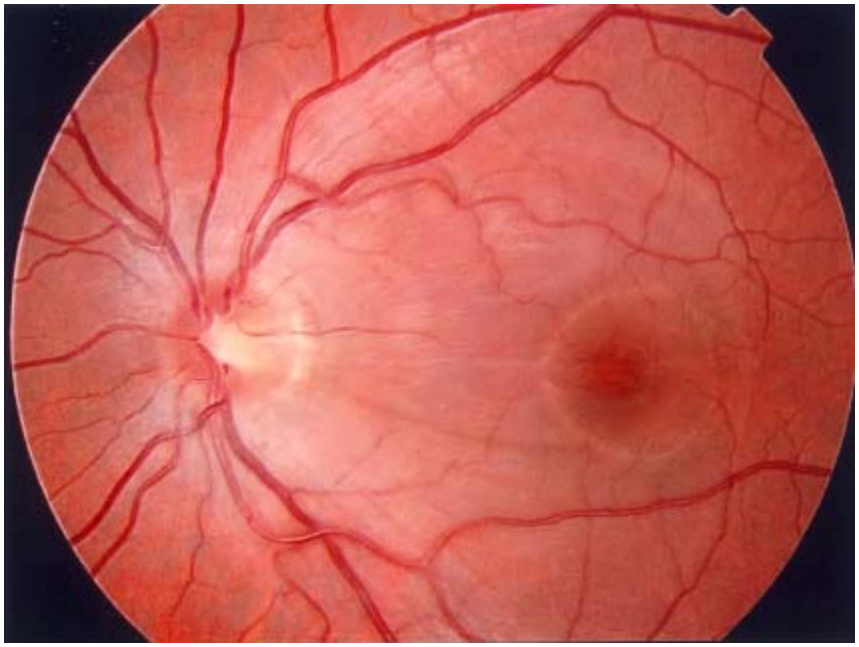

FIGURE 1C

Case 2. Dynamic atypical optic nerve coloboma with transient macular detachment in 30-year-old man with a 1-week history of blurred vision in the left eye. Fundus photograph of the left eye shows an apparently normal nerve with a small slit-like cup and associated macular schisis with outer layer macular detachment. Visual acuity was 20/160.

Observation was recommended, and 8 months later the uncorrected acuity improved to 20/20. The schisis cavity and outer layer detachment were significantly diminished, yet the optic nerve now contained a temporal excavation (Figure 1D). Late-frame disc hyperfluorescence was no longer present by angiography. Funduscopic examination of the right eye was unchanged throughout the course of observation. The funduscopic appearance could not be reproduced by changes in body position or by light stimulation of the eye. OCT was unavailable.

Case 3

A 19-year-old white woman presented with blurred vision in the right eye that had been present for 6 months. Best-corrected acuity was 20/60 in the right eye and 20/25 in the left eye. Macular pigmentary abnormalities suggestive of previous macular schisis and optic nerve excavations suggestive of either large temporal optic pits or atypical colobomas were noted in both eyes (Figure 1E). Both optic nerves were normal in size and did not have any peripapillary pigmentary changes. We could not identify a macular schisis or an outer layer detachment in either eye. Five months later, this patient was examined urgently for an acute drop in visual acuity to 20/60 in the left eye. We observed a pronounced change in the funduscopic appearance of the left eye. The optic nerve now had a slit-like cup, and the macula contained a large schisis cavity with a centrally located outer layer detachment (Figure 1F). We also noted a macular schisis without an associated outer layer detachment in the right eye.

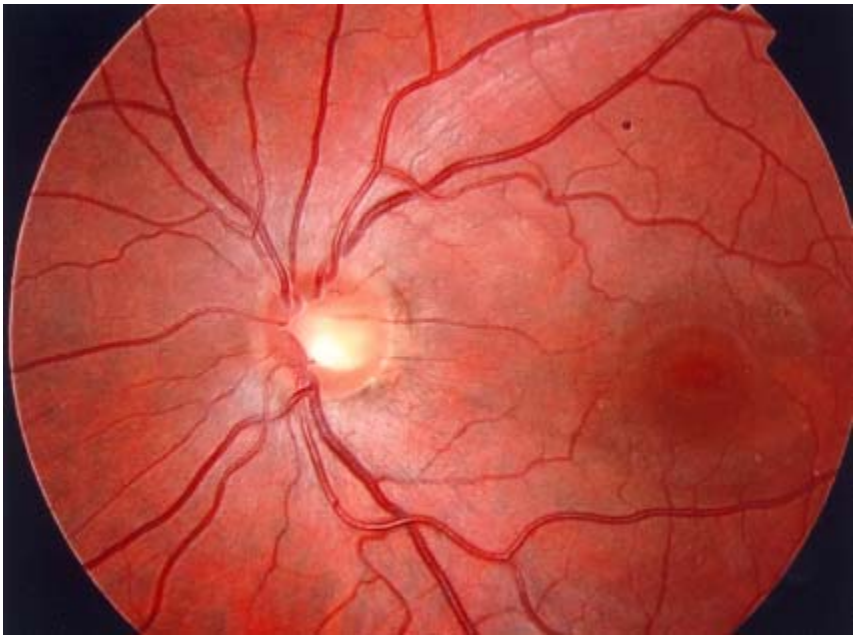

FIGURE 1D

Case 2, left eye, 8 months after photograph in Figure 1C was taken. No treatment had been administered. An optic nerve cavitation is present associated with marked improvement of macular schisis and outer layer macular detachment. Visual acuity improved to 20/20.

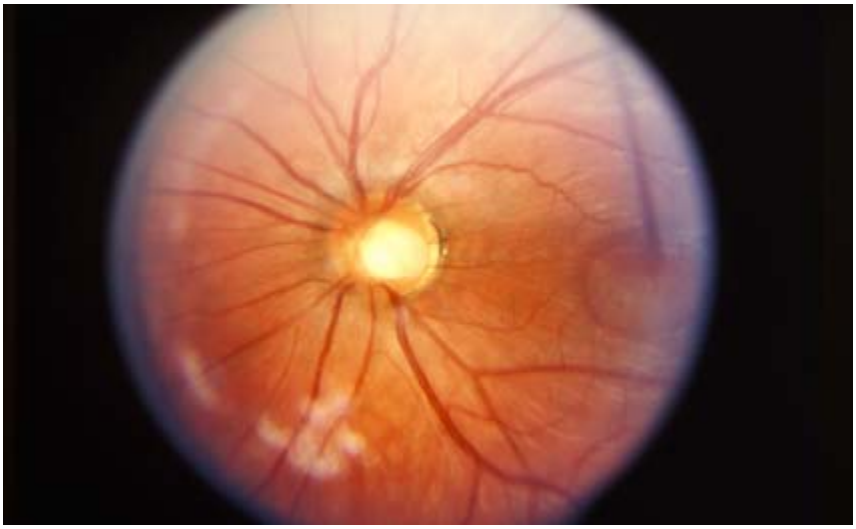

FIGURE 1E

Case 3. Dynamic atypical optic nerve coloboma with transient macular detachment in 19-year-old woman. Fundus photograph of the left eye shows initial appearance of atypical optic nerve coloboma. Macular pigmentary changes are present without macular schisis or detachment. Visual acuity was 20/25.

We could not detect an alteration in the right optic disc, nor could we find an outer layer hole or a posterior vitreous detachment in either eye. Both eyes had late disc hyperfluorescence on angiography.

We recommended observation, and over the next 5 months acuity improved to 20/40 in the left eye and remained 20/60 in the right eye. The macular schisis began to flatten bilaterally, but foveal retinal pigment epithelial changes were still noted bilaterally. A dramatic change in the appearance of the left optic nerve was seen, which was noted as early as 1 month after initial observation of the

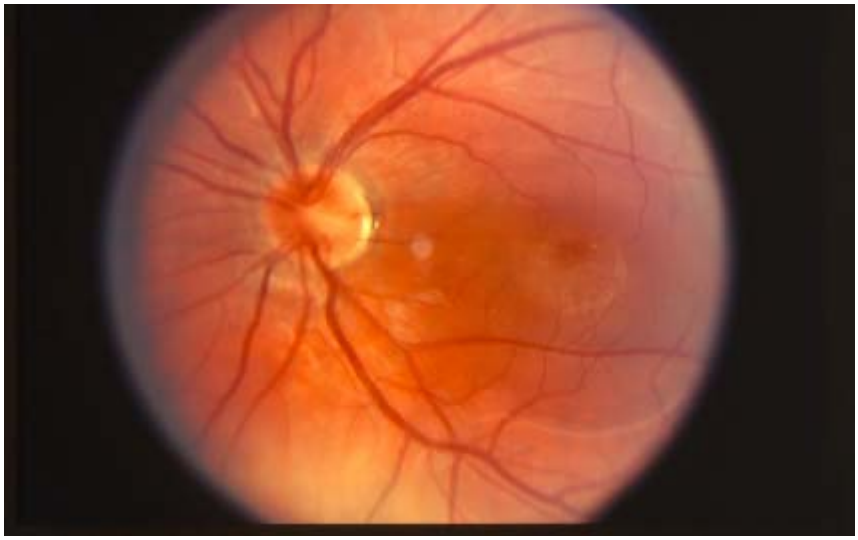

FIGURE 1F

Case 3, left eye, 5 months after photograph in Figure 1E was taken. An apparently normal nerve with slit-like cup and associated macular schisis and outer layer macular detachment are present. The previously documented atypical optic nerve coloboma was not visualized. Visual acuity was 20/60. Brightness and contrast of image have been modified.

slit-like cup. The temporal optic nerve excavation originally noted had reappeared (Figure 1G). The right optic nerve was unchanged. We uncovered no systemic associations. The patient remained normotensive, and the funduscopic appearance could not be reproduced or altered with changes in body position or by light stimulation. Continued observation was recommended. OCT was unavailable.

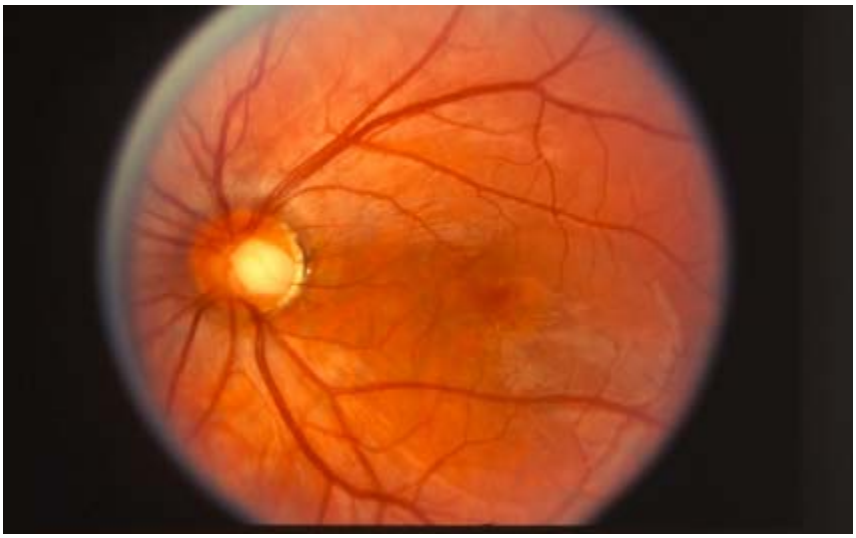

FIGURE 1G

Case 3, left eye, 5 months after photograph in Figure 1F was taken. No treatment had been administered. The atypical optic nerve coloboma had reappeared, and the macular detachment had diminished. Retinal pigment epithelial changes were noted in the macula. Visual acuity was 20/40. Brightness and contrast of image have been modified.

DISCUSSION

Optic nerve pits and typical colobomas are probably embryologically distinct congenital excavations of the optic nerve.[2,4] Whereas typical colobomas are located inferonasally, pits are usually small, well-demarcated, temporally located depressions in the nerve substance. Pits are sometimes even categorized as a subset of atypical optic nerve colobomas.[4] The optic disc anomalies in our patients are similar to those reported by Johnson and Johnson[4] as well as observed in an autosomal dominant pedigree of atypical optic

nerve colobomas and pits that were associated with macular detachments.[14] Our patients lacked a family history of optic nerve disease.

In this series, we report the development of macular schisis in four eyes of three patients. Unlike the macular schisis typically associated with cavitary optic nerve anomalies, in at least one eye of each of these three patients, an optic nerve excavation was definitely not seen at the onset of the macular schisis. As the macular schisis resolved in our three cases, simultaneous, ipsilateral optic nerve changes occurred. In two of the cases, only then did it become apparent that these eyes contained optic nerve excavations. In the third case, the patient was examined prior to the development of the macular schisis, at which time a cavitary anomaly was observed. In this case, we documented the disappearance of the excavation along with the coinciding development of macular schisis. Subsequently, as the excavation reappeared, the macular schisis resolved. The third case is also unusual in that bilateral macular changes occurred simultaneously. In all cases, the optic nerve and macular changes occurred over several months, and in no case could the changes be reproduced by external stimulation.

Though earlier reports described the association of macular schisis and optic pits,[1,5] Lincoff and associates[15] were the first to detail the macular changes in this condition and develop a unifying hypothesis regarding the pathophysiology. Since the publication of this seminal article, reports describing the OCT findings of optic pit maculopathy have confirmed Lincoff's hypothesis.[16,17] The macular changes in our patients were identical to those described in these reports. All of our patients had evidence of a macular schisis, and in three of the four eyes, an outer layer detachment was identified. In no case was an outer layer hole seen. Unfortunately, OCT was not readily available when we examined these patients, but it perhaps would have given more insight into our observations.

Macular changes have been reported with many disc anomalies, including optic nerve pits,[1,5] typical and atypical optic disc colobomas,[4,18] and morning glory syndrome.[13] Spaide and associates[19] have even reported the clinical and OCT findings in a patient with macular schisis without an optic disc pit. Our cases are similar to the one reported by these investigators in that when our patients presented with macular schisis, a cavitary optic nerve anomaly was not visible. However, unlike our three cases, Spaide and associates[19] did not observe any optic nerve changes in their patient through 4 years of follow-up. Additionally, Walsh and Hoyt[20] described a 14-year-old white girl with central serous retinopathy in which an optic pit was not seen during the first year of observation but later became apparent. Over a 9-year period, the investigators did not document a fluctuation in the optic nerve appearance despite noting spontaneous clearing of the central serous retinopathy on two occasions. In our cases, the optic nerve and macular changes were documented over a 5- to 8-month period. Since patient 3 was under our care prior to the development of the optic nerve and macular changes, we were able to observe changes in the optic nerve that may not otherwise have been suspected. Despite the noted differences, it is possible that the cases reported by Walsh and Spaide represent a variant of the condition we are herein reporting.

Moore and associates[21] reported a patient with autosomal dominant atypical optic nerve coloboma that developed progressive worsening of optic nerve excavation over a period of 13 years, unassociated with serous macular detachment. Worsening was attributed to low-tension glaucoma. Our cases appear to represent a different, more acute process because of the short time period over which disc excavation appeared (1 to 8 months), initial appearance of disc hyperfluorescence on fluorescein angiography, and reversal of associated serous macular detachment. Case 3 also showed reappearance of a previously documented disc excavation, suggesting that the excavation constituted a baseline abnormality not attributable to glaucoma during the observation interval.

The fluctuating appearance of peripapillary staphylomas,[11] optic nerve colobomas,[12] and morning glory discs[13] has previously been reported. In all reported cases, these changes typically occur over a several-minute period instead of a several-month period as we report. In addition, to our knowledge, concurrent macular changes have not been previously observed except in association with the morning glory disc anomaly.[13] Our cases lacked features typical of morning glory disc anomalies, peripapillary staphylomas, or typical optic disc colobomas. Given the normal size and appearance of the nerve and associated vessels and peripapillary retina, our cases most likely represent atypical optic nerve colobomas.

The spontaneous resolution of the macular schisis in our cases is not unique; this phenomenon has been reported infrequently.[3,5,10] In fact, many investigators favor treatment of macular schisis associated with optic nerve pits and other disc anomalies because of the poor natural history.[9,10,22] However, the unusual presentation of our patients and the variable response to treatment reported in the literature led us to recommend observation in these cases.[9,22] It is uncertain whether our patients would have had better visual outcomes or quicker visual recovery had a surgical procedure been performed.

Both the source of fluid and the underlying mechanism responsible for the development of optic pit maculopathy remain elusive.[4-9] Though other investigators have postulated that forces along the vitreoretinal interface are responsible for optic pit maculopathy,[23] we were unable to convince ourselves that these forces were responsible for the macular and optic disc changes seen in our patients. Additionally, the development of bilateral simultaneous macular schisis in our third patient at least suggests a systemic influence despite our inability to determine one.

Cavitary optic nerve anomalies are herniations of dysplastic retina through a defect in the lamina cribrosa, in which tissue sometimes even extends posteriorly into the subarachnoid space.[5,7,16,24] Even though histologic evidence for a direct communication between the pit and subarachnoid space is lacking,[5,24] experimental and some clinical evidence suggests it is conceivable.[7,8] Alternatively, there is compelling clinical evidence that in some cases a direct connection exists between the vitreous and subretinal space via a defect in the excavation.[4,9] Recently, Johnson and Johnson[4] proposed that the excavation could act as a bulb syringe and subsequent fluctuations in the intracranial pressure could lead to the development of macular schisis. We believe that this hypothesis can explain the optic nerve and macular changes noted in our three patients. Cavitary anomalies likely contain dysplastic tissue that is multilaminar and in some instances porous, which could provide interconnections between the vitreous, the subretinal space, and

possibly the subarachnoid space (Figure 2A). These features would allow the anomaly to act as a bulb syringe. As the formed vitreous liquefies with age, some fluid could migrate through a tissue defect, possibly as a result of intracranial pressure fluctuations. At a later time, a sudden or pronounced increase in intracranial pressure could force the liquid vitreous anteriorly from a sac in the cavitation into the retinal tissue, thus creating a schisis. If some of this fluid became trapped under redundant dysplastic tissue normally lining the anomalous wall, anterior displacement of this tissue could change the appearance of the optic nerve (Figure 2B). We believe this concept explains the observations in our three patients. It is interesting that in our third patient, optic nerve changes were seen in only one eye. We suspect that the constellation of histologic features necessary for our observations is rarely present, perhaps further explaining why these findings have not previously been reported. Diagnostic technology like OCT might be helpful in confirming or refuting this hypothesis.

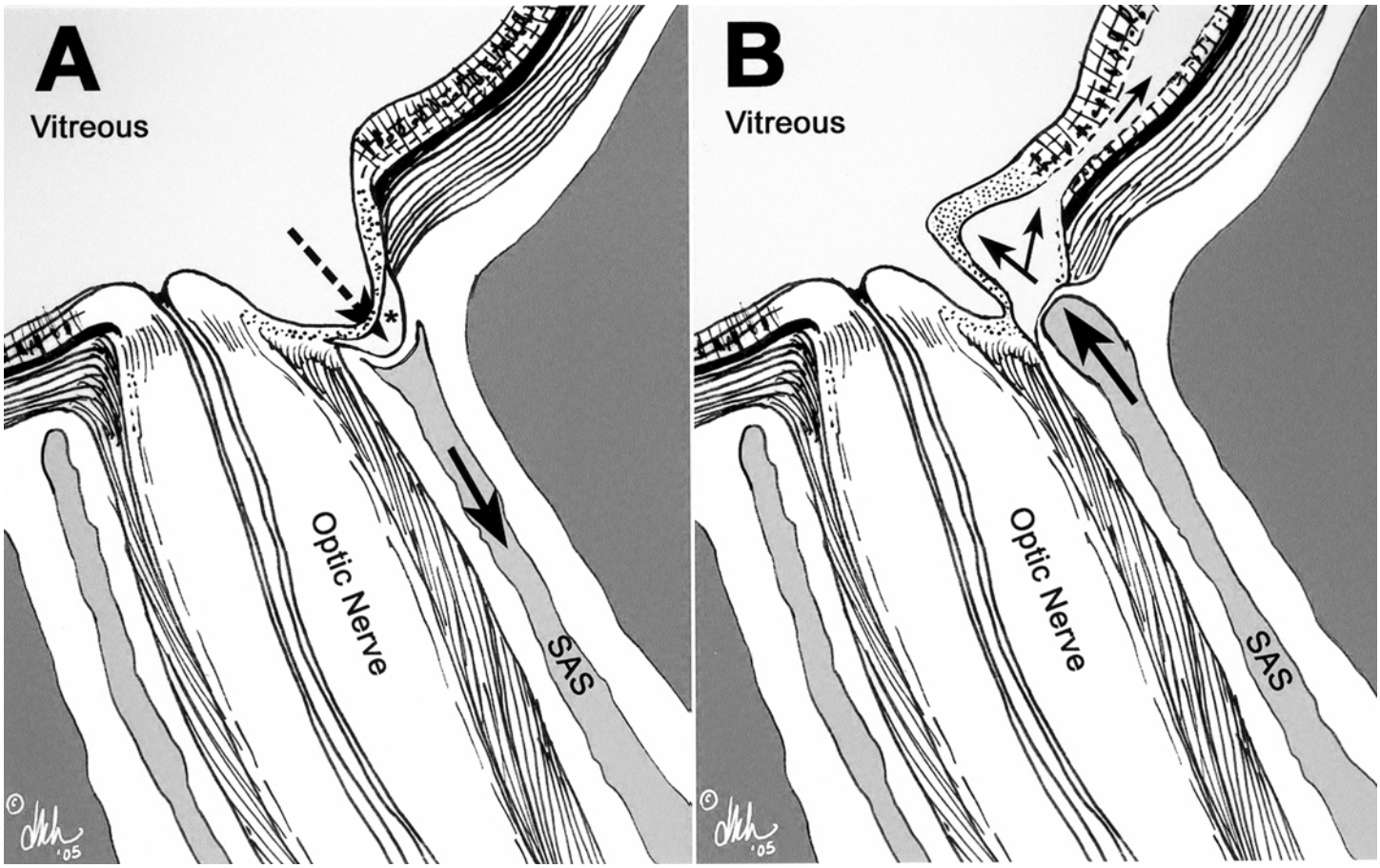

FIGURE 2

Cross-sectional schematic of optic nerve in case of dynamic atypical optic nerve coloboma with associated maculopathy. It is hypothesized that herniation of variably porous dysplastic tissue and its proximity to the subarachnoid space (SAS) could allow liquefied vitreous to migrate posteriorly into a sac* with fluctuations in intracranial pressure (A). Later, increases in intracranial pressure could force fluid anteriorly, resulting in schisis formation and the displacement of nerve tissue, thus altering the optic nerve appearance (B).

In summary, we present three patients with macular schisis and concomitant optic nerve changes. Though unproven, these changes could be the result of fluctuations in the pressure gradient between the subarachnoid space and the vitreous cavity. In particular, the simultaneous development of bilateral macular detachments in our third patient lends further evidence that systemic factors may play a role in the development of macular changes in patients with cavitary optic nerve anomalies. Clinicians should be aware that in cases of macular schisis in which an optic nerve excavation is not initially apparent, spontaneous resolution of the macular schisis is possible. Concurrent optic nerve changes should be sought and in some cases will reveal a cavitary anomaly. In the future, advances in imaging technology may allow further understanding of the pathophysiology in this unusual condition.

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PEER DISCUSSION

DR MORTON S. COX. I congratulate Dr. Han and his colleagues for their observations. I found the same phenomenon when I reviewed one of my similar cases. Little or no cavitation of the right disc compared to the left was present at the onset of the macular detachment in the right eye. Disc cavitation appeared as the maculapathy resolved following laser photocoagulation of the temporal juxtapapillary retina. When the left macula detached acutely three months later the previously noted coloboma was obscured only to reappear as the maculopathy improved after treatment. However, this phenomenon was not always present. The coloboma persisted in spite of the macular detachment in a second case of mine and in the right eye of the author's third case.

Perhaps inflammation and swelling obscures or contributes to the obscuration of the coloboma in some of these cases. The disc is hyperemic and the authors report late papillary and juxtapapillary hyperfluoresence that is not usually associated with optic nerve pit maculopathy. The acute onset (overnight in one of my cases) of a relatively large macular schisis and detachment may have insulted the integrity of the vascular compartment causing a secondary swelling of the disc.

The apparent cavitation that accompanies the resolution of these macular detachments should not be confused with the more serious often overlooked progressive cupping and loss of nerve tissue that occurs over time in some of these cases due to glaucoma or low tension glaucoma. Savell and Cook[1] reported a greater incidence of total cupping in the older compared to the younger members of a family with an autosomal dominant pedigree. The apparent loss of tissue from the colobomatous nerve with increasing age was attributed to an increased susceptibility to the intraocular pressure. Moore and coworkers[2] documented progressive optic nerve cupping, loss of neural rim tissue, and thinning of the retinal nerve fiber layer over l3 years in a patient with familial, bilateral, atypical optic nerve colobomas and normal intraocular pressure. The left eye of the author's first case was cupped due to low-tension glaucoma and the coloboma of the left eye of one of my patients increased to total cupping over 10 years due to known open angle

glaucoma in spite of treatment. Regardless of the status of the macula these patients need regular examinations with careful documentation of the optic disc and visual fields because of their increased sensitivity to the intraocular pressure. Family members deserve the same attention because of the frequent familial occurrence. Perhaps progressive cupping is an indication to lower the intraocular pressure regardless of it's level although the efficacy of such treatment is unknown.

Over five to eight months the authors observed improvement of the vision and maculas without treatment in all of their cases. However, macular pigmentation was also noted in all cases and its presence initially in the right eye of the author's third case indicates an earlier detachment. Spontaneous reattachment is common over the long run but the vision is usually poor due to long standing detachment or multiple recurrences as noted in the left eye of one of my patients with 20/400 visual acuity after l0 years observation. There was marked destruction of the retinal pigment epithelium in the area of the previous outer layer detachment. In my opinion it is unlikely that the vision and maculas of the author's patients will remain stable indefinitely without treatment.

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DR IRENE H. MAUMENEE. The authors describe a patient with findings very similar to those of a patient that Dr. Cameron Parsa and I described at the Meeting of the American Ophthalmological Society in 1998[1]. The patient had been treated for many years with a diagnosis of low-tension glaucoma, even though he gave a history compatible with bilateral recurrent exudative detachments. His optic nerve appearance showed a congenital malformation of the nerve head. On Doppler ultrasound we could not demonstrate a central retinal artery. The optic nerve head showed an excess of blood vessels extending over the rim of the optic nerve; the posterior ciliary artery presumably perfused these vessels. The patient also developed progressive renal failure. This entity is autosomal dominant and commonly referred to as papillorenal syndrome. You may want to check your patient for proteinuria and creatinine and, using ultrasound, for size of his kidneys.

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DR LEE M. JAMPOL. Only one of three of your patients seems to be dynamic; the other two just showed progressive cupping. I concur with Dr Mort Cox's comment that you have to be certain that was not an irreversible cupping that occurred in those cases in response to low-tension glaucoma. The Moore and colleagues paper that was cited by both you and Dr Cox is part of a large pedigree of patients with this disease, with a very variable expressivity. I think we have 18 involved patients and three of them show progressive, apparently irreversible, cupping in the optic nerve despite normal intraocular pressures. This may be a type of low-tension glaucoma in an anomalous nerve, and two of your three cases might represent that entity.

We have four patients that have glaucomatous cupping and have developed, without a pit or any previously known disease, a schisis of the macular area. I think that the schisis phenomenon can occur not only with pits and colobomas but can occur from acquired glaucomatous cupping.

DR ELIAS I. TRABOULSI. I have a question about the terminology. These patients have optic pits and, to call them colobomas, seems like you are just trying to avoid calling them pits. They are not the most typical pits but your OCT findings are very compatible as is the clinical picture. The interesting finding is that they have been bilateral. I tend to reserve the term coloboma to the more typical coloboma – inferior failure of closure of the fetal fissure etc. Dr Maumenee was referring to the *PAX2* related atypical coloboma where the excavation is throughout the disk; these are the patients who have renal malformations and so on. We had a family with this clinical presentation as you have shown and did not find a PAX2 mutation. I suggest evaluating the renal function in these patients anyway. Your observation of the variation in the size of that pit relates to the fact that you had multiple occasions to see them when there was fluid or where there was no fluid. I would still call them straightforward optic pits. Cavitary malformation is also appropriate because it's a little bit broader, but the OCT findings and everything else is compatible with a pit.

DR GARY C. BROWN. Determining whether it is a pit or a coloboma may be a matter of definition. Wise described pits in about 1883; so it took 122 years for people to further define the condition. When the people develop a retinal detachment, the visual prognosis is not good. Both the Iowa series[1] and our series[2] show that over 50% of the people are close to legal blindness or legally blind at the end of one year with no treatment. About 25% of the people who develop a retinal detachment will also develop an outer layer macular hole which happens fairly early, even within a couple of weeks. Once they develop that hole, in our experience, they just don't do well with any type of treatment. You might consider the application of peripapillary laser therapy, which has low morbidity early on.

REFERENCES

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2. Brown GC, Shields JA, Goldberg RE. Congenital pits of the optic nerve head. II. Clinical studies in humans. *Ophthalmology* 1980;87:51-65

DR LEE M. JAMPOL. I would not call these optic nerve pits. If you do then you will not be communicating effectively with retinal specialists and other ophthalmologists. I don't think this is what most ophthalmologists consider to be an optic nerve pit. I like the term, "atypical coloboma."

DR DENNIS P HAN: We did not perform any evaluation for renal evaluation or evaluation of the PAX-2 status in these patients. Are these cases demonstrating progressive cupping or, alternatively, obscuration of a pre-existent disc anomaly that returned to baseline when the macular fluid resorbed? The latter is supported by the fact that we have some other anomalies demonstrated on fundus fluorescein angiography that are more of an acute nature; these would not usually be observed in cases of progressive loss of nerve tissue due to low-tension glaucoma. Also, in the reports of glaucoma or low-tension glaucoma in the literature, the progressive cupping occurred over a long period of time. Moore reported a case with progressive cupping that occurred over a period of 13 years as opposed to the cases where we have shown that the appearance of disc excavation has occurred over a matter of months. The fact that macular detachment schisis can occur in low tension or acquired glaucomatous cupping is very interesting.

We struggled with the terminology of the disc anomaly. When reviewing the literature, many authors preferred not to describe these kinds of anomalies as pits, because the term "pit" has typically been used to describe a small focal oval defect within a larger excavation in the optic nerve that may have a greenish-gray coloration. That being said, there are families that have anomalous excavation and optic pits within the same pedigree. Dr Madison Slusher published a paper years ago[1] suggesting that these may represent variations of the same condition. Our bias is to describe this as an atypical optic disc coloboma to be more consistent with the previous literature.

In terms of treatment, there appears to be some type of acute phenomenon that may be different from other serous maculopathies that occur with pits or colobomas as evidenced by the presence of the fluorescein leakage, the relatively acute onset, and the hyperemic disc, which are not typically observed. Because the pathophysiologic process may be different, a period of observation may be worthwhile. Our three cases all had a good visual outcome. However I concede that at some point, especially if serous macular detachment recurs or if there is a prolonged course, intervention should be considered.

REFERENCE

1. Slusher MM, Weaver RG, Greven CM, et al. The spectrum of cavitary optic disc anomalies in a family. *Ophthalmology*. 1989;96:342-347.