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## Optic Disc Pits: A Case Report and Review

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## Optic Disc Pits: A Case Report and Review

### Abstract

**Background:** Congenital optic disc pits (ODP) are a rare clinical finding affecting approximately 1 in 11,000 people. Affected individuals are generally asymptomatic unless fluid accumulates in the macula resulting in severe vision loss. The management of ODPs depends mostly on clinical findings and can range from observation to surgery. Optometrists need to be aware of clinical presentations and possible complications of ODPs.

**Case Report:** This report will review a case of an asymptomatic optic disc pit and discuss the potential treatment options if complications arise.

**Conclusion:** Although rare, congenital optic disc pits need to be accurately assessed and diagnosed. Symptoms can range from none to severe vision loss. Proper observation and management may lead to improved visual outcomes.

### Keywords

Optic disc pit (ODP), maculopathy, ocular coherence tomography (OCT), retinoschisis

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## INTRODUCTION

The evaluation of the optic nerve is a critical part of any optometric examination. These nerves can have a “normal” appearance that varies widely in size, shape, and color. Atypical nerve head appearances can be due to congenital anomalies or pathological changes. So, it is important for the clinician to be able to distinguish normal versus abnormal and to identify the correct etiology.

Congenital optic disc pits (ODP) are a rare clinical finding affecting approximately 1 in 11,000 people.<sup>1,2</sup> Affected individuals are generally asymptomatic unless fluid accumulates in the macula resulting in severe vision loss.<sup>3</sup> The management of ODPs depends mostly on clinical findings and can range from observation to surgery. Optometrists need to be aware of the clinical presentations and possible complications of ODPs. This report will review a case of an asymptomatic pit and discuss the potential treatment options if complications arise.

## CASE REPORT

A 56-year-old African American female initially reported to our clinic for a comprehensive eye examination. She had no visual or ocular complaints and wanted to update her spectacle correction. Her medical history was positive for systemic hypertension controlled with hydrochlorothiazide 25 mg tablets once daily and acid reflux for which she was taking famotidine 20 mg once a day. She reported that she “has something in the inside of one of her eyes” but does not recall more information than that. She denies any other significant personal or familial ocular history.

Her uncorrected vision was finger counting at 5 feet in both eyes. A manifest refraction of -6.25-1.25 x 170 OD and -5.00-0.50 x 020 OS corrected her to 20/25 OD and 20/20 OS. Her extraocular muscles were unrestricted. Pupils were equal, round, and reactive without an afferent pupillary defect. Confrontation visual fields were restricted in the superior nasal quadrant of the right eye; the other quadrants were full, as was the left eye. Intraocular pressures measured 14 mm Hg in each eye by Goldmann applanation tonometry.

A slit lamp exam of her anterior segment revealed normal eyelids. Corneas were clear and her angles were open. The anterior chambers were deep and free from cell and flare. Her irises were brown and flat. She was dilated with one drop each of phenylephrine 2.5% and tropicamide 1% in both eyes. The lenses of each eye

showed early nuclear sclerosis, and the vitreous was clear with no evidence of posterior detachments. The cup to disc ratio of the right optic nerve was estimated at 0.70. The nasal rim tissue appeared healthy; however, the rim was significantly thinned from 6 to 9 o'clock and the disc was tilted temporally. An area of retinal pigmented epithelium (RPE) hyperplasia was seen adjacent to the thinned rim tissue. Additionally, a circular gray area of excavation was observed on the temporal side of the right optic cup. The left optic nerve had an intact neuro-retinal rim tissue that was normal in color, and no excavations were seen. Its cup to disc ratio was 0.50, and a scleral crescent was noted on the temporal side of the nerve (Figure 1). The maculae were flat in each eye. The vessels were of normal course and caliber, and the peripheral retina of each eye was intact without any evidence of holes, tears, or detachments.

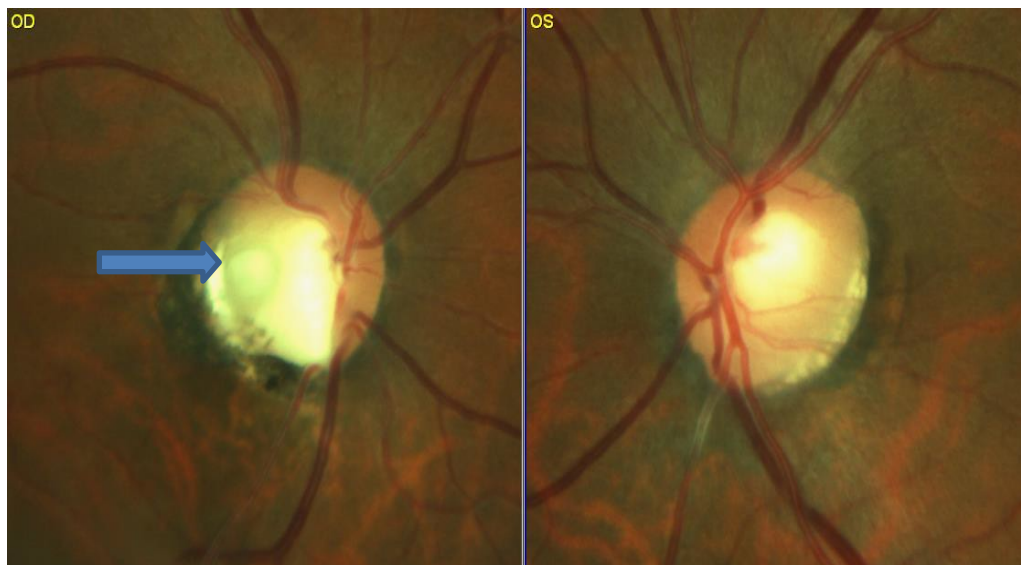


Figure 1 – Photographic comparison of right and left optic nerves. The excavated area (blue arrow) is seen temporally in the right nerve.

To assist in the diagnosis of this patient, optical coherence tomography (OCT) scans of the macula and retinal nerve fiber layer (rNFL) were obtained. The scans showed a standard foveal contour in each eye. Significant thinning from 6 to 8 o'clock was seen on the rNFL scan of the right eye, and the area of excavation can be seen temporally on the vertical tomogram. The scan of the left eye showed minimal focal thinning inferiorly (Figure 2).

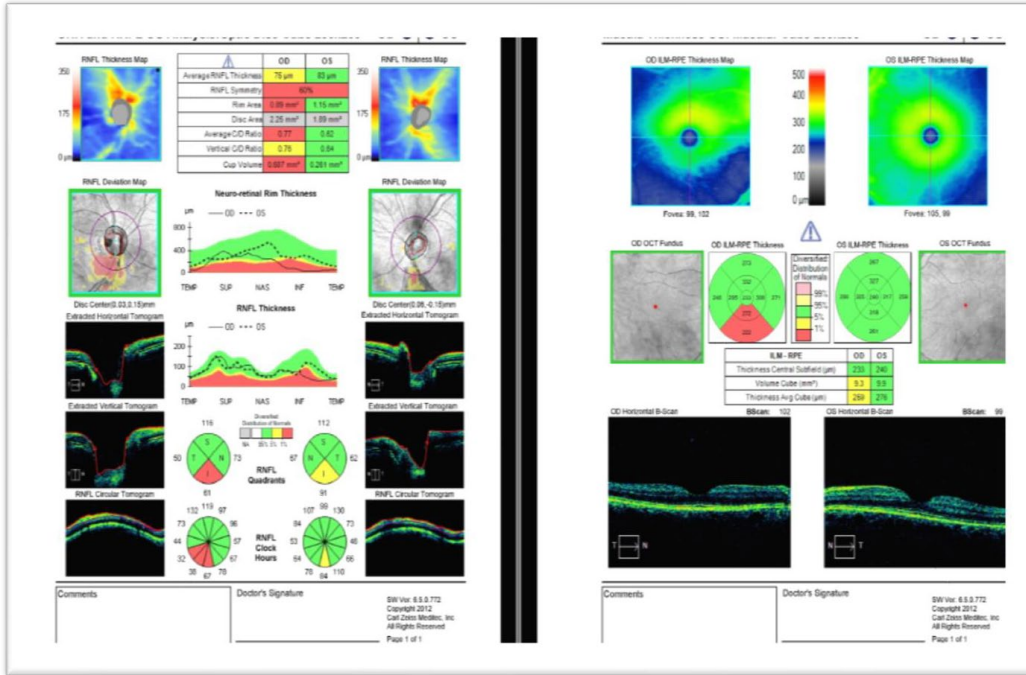


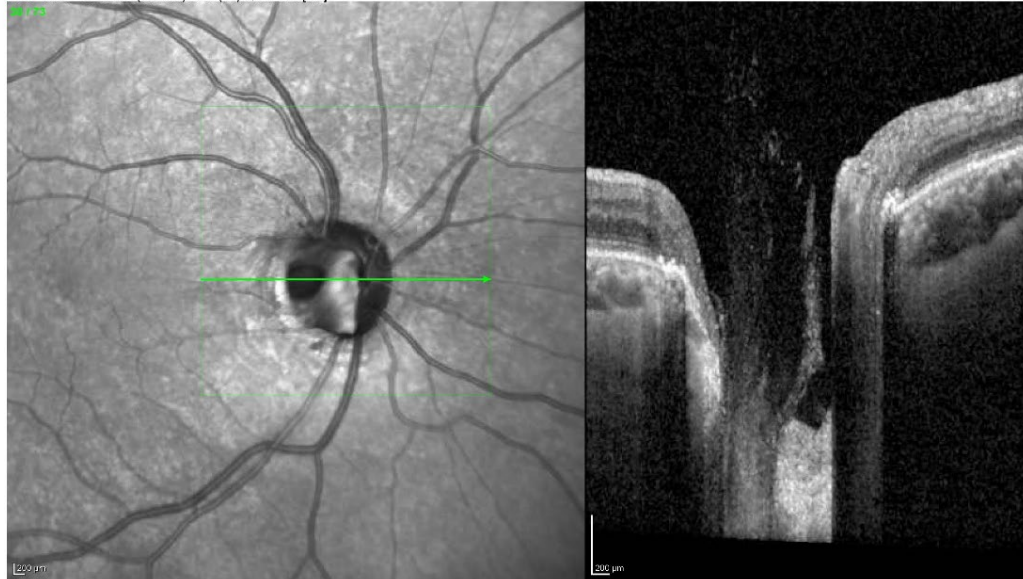
Figure 2 - rNFL and Macular Cube scans of each eye.

**Overview Report**  
SPECTRALIS® Tracking Laser Tomography

Patient: \_\_\_\_\_ DOB: \_\_\_\_\_ Sex: F  
Patient ID: \_\_\_\_\_ Exam.: \_\_\_\_\_  
Diagnosis: --- Comment: ---

**HEIDELBERG ENGINEERING**  
**OD**

IR 30° ART + OCT 15° (4.8 mm) ART (11) Q: 28 EDI [HS]



Notes: \_\_\_\_\_  
Date: 2/25/2020 Signature: \_\_\_\_\_

Figure 3. Scan through the optic pit. Pit appears to be filled with vitreous.

After the fundoscopic, photographic, and tomographic evaluation, EB was diagnosed with a congenital optic pit of the right eye along with sclerotic nuclear cataracts and compound myopic astigmatism OU. She was asked to return at her earliest convenience for threshold visual field testing. Due to the risk of a serous retinal detachment, a home Amsler grid was dispensed to the patient along with instructions to return to the clinic as soon as possible if changes were noted.

FOLLOW UP #1

EB returned a week later for visual field testing. She had no new ocular or visual complaints. Her corrected visual acuities were stable at 20/25 OD and 20/20 OS. Her anterior segments were unchanged, and intraocular pressures were 15 mm Hg OD and 14 mm Hg OS by Goldmann applanation tonometry. Pupils were equal in size, round, and reactive. A central 24-2 Threshold Test was performed on each eye using a Humphrey Visual Field Analyzer II (Figure 3). The right eye's field showed a defect in the superior half of her vision that was denser nasally; the left eye's visual field was within normal limits. EB was counseled with regard to her visual field results and asked to return in six months to be re-evaluated.

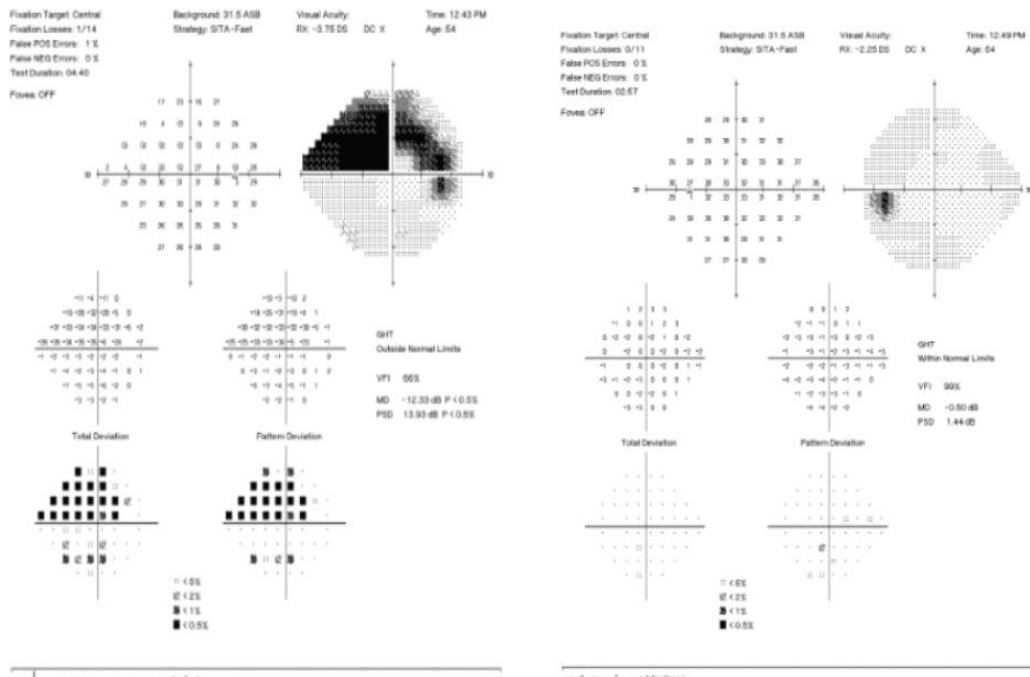


Figure 4 Visual Field of right (left image) and left eye (right image)

FOLLOW-UP #2

The patient returned in six months for a follow-up and reported no new symptoms or complaints. Corrected visual acuities remained at 20/25 OD and 20/20 OS. Goldmann applanation tonometry measurements were 14 mm Hg in each eye. The anterior segments were unremarkable for any changes, and pupillary responses were normal. EB was again dilated with phenylephrine 2.5% and tropicamide 1% to visualize the posterior segment. The sclerotic nuclear changes of her lenses showed no significant progression. Her optic nerve heads were also unchanged in appearance. The maculae and peripheral retina of both eyes were flat and intact. A visual field test (Figure 4) and a rNFL scan (Figure 5) were repeated.

Once again, the left visual field was normal. The right eye had a stable, superior arcuate defect that correlated with the nerve fiber layer loss seen in the original (Figure 2) and secondary (Figure 6) OCT. There was no clinically significant progression in the visual field defect or nerve fiber layer loss which supported the diagnosis of a congenital optic pit versus a glaucoma-related acquired optic disc pit.

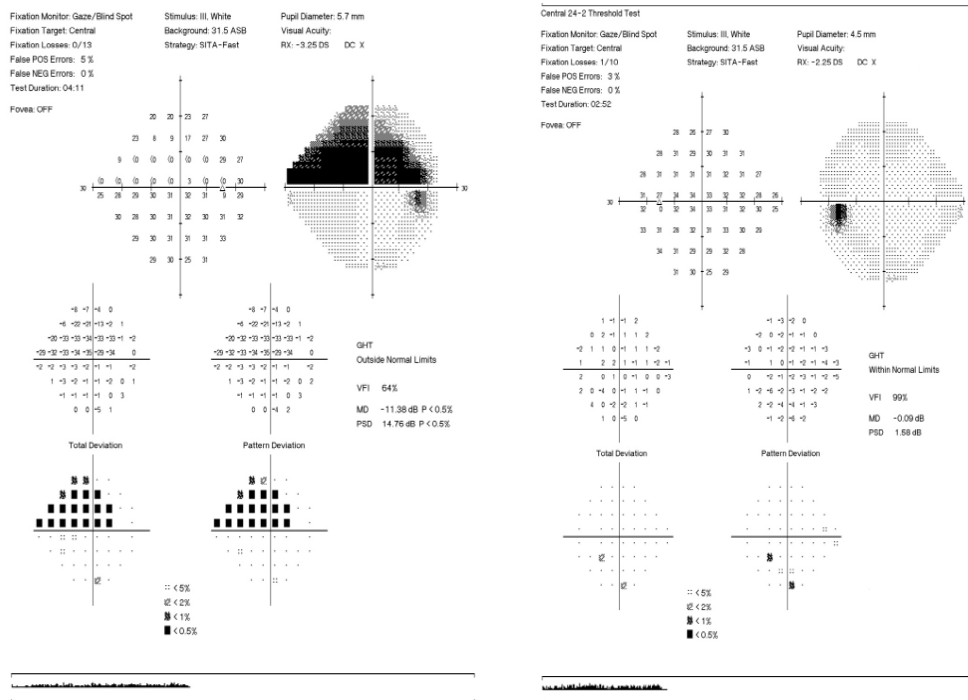


Figure 5 – Repeat testing of the patient’s visual field shows no signs of progression.



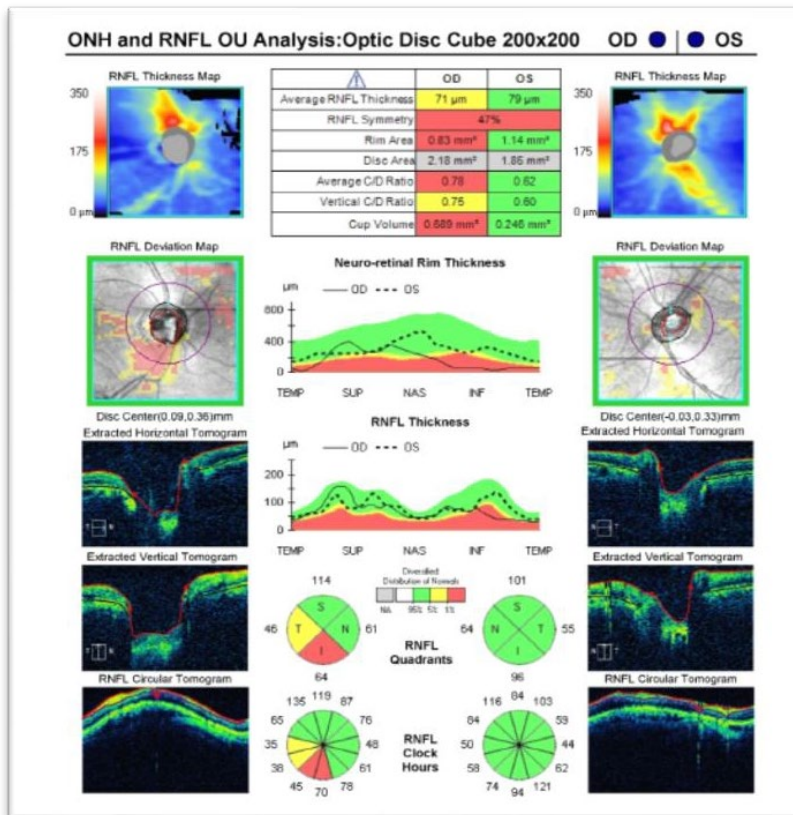


Figure 6 Repeat of the rNFL of both eyes. The rNFL loss was stable in the right eye

All findings were reviewed with the patient, and she agreed to be seen every six months for a dilated fundus exam and visual field testing.

## DISCUSSION

The differential diagnoses for anomalous optic nerves may include glaucoma, hypoplastic discs, optic nerve colobomas, morning glory syndrome, and tilted/malinserted discs. In this case, it was difficult to rule out glaucoma as a comorbidity. However, with an essentially normal rNFL in the left eye and normal intra-ocular pressures, this patient was not believed to have glaucoma but continues to be monitored for changes in her optic nerve cupping, nerve fiber layer, and visual field defects. Also, this patient was not noted to have a relative afferent pupillary defect. This is an unusual finding, or lack of finding, considering her visual field defect in the right eye. This may be a true anomalous finding, but human error in detecting a subtle APD is possible.

Since the right optic disc was larger than the left optic disc, along with the absence of the classic “double-ring sign,” optic disc hypoplasia was ruled out. This case also



lacked the typical optic nerve head coloboma appearance of a large, well-delineated white area of excavation. The margins of both discs were flat, ergo tilted and malinserted discs were both ruled out; however, the patient did have a scleral crescent around the left optic disc.

Congenital optic disc pits are malformations of the optic nerve and are usually categorized with other abnormalities such as optic nerve head colobomas, morning glory, and tilted discs. They are a relatively rare finding that only occurs in about 1 in 11,000 people.<sup>1,2</sup> ODPs usually present unilaterally, although nearly 15% of cases present bilaterally. Their clinical presentation is variable, but most pits are located in the inferior-temporal section of the nerve head.<sup>4,5</sup> The color of an ODP may depend on the amount of glial tissue within the pit. While most are gray in appearance, they may also appear white, yellow, or black.<sup>4</sup> The size of ODPs is also variable. A 2013 report from Japan measured 15 ODPs and found the average diameter to be 490.4  $\mu\text{m}$  or roughly 0.32-disc diameters; however, size can range from 0.1 to 0.7-disc diameters.<sup>8</sup>

The origin of ODPs is an area of debate. For many years congenital optic disc pits were believed to occur if the embryonic fissure failed to close normally in the second month of gestation, as in the case of colobomas.<sup>4,6,7</sup> However, two points have been used to challenge this convention. The first consideration is that if colobomas and ODPs shared the same origin, it would stand to reason that they would often appear in the same eye, but they do not.<sup>1,4</sup> Secondly, ODPs are not associated with systemic disease, but up to two-thirds of patients with a coloboma have a non-ocular disorder.<sup>6,7</sup>

One alternate theory suggests that ODPs are formed in the fifth month of gestation rather than the second. It is during the fifth month when mesodermal cells move from the sclera to a location that eventually becomes the lamina cribrosa. It is theorized that poor migration and differentiation of these mesodermal cells leads to the formation of an optic disc pit.<sup>8</sup> This theory is supported by OCT and histological studies that have shown ODPs to be areas of dysplastic retina tissue that have herniated through a defect in the lamina cribrosa.<sup>4,9</sup>

Clinically, ODP patients are often asymptomatic. However, they may have reduced visual acuity and/or visual field defects due to displacement or thinning of the retinal nerve fiber layer at the site of the ODP.<sup>10</sup> A superior arcuate scotoma, like the one seen in our case, is the most common field defect but other field defects are possible as well. Field deficits associated with ODPs do not typically progress.<sup>9</sup>

Along with the visual defects caused by an ODP, affected patients may develop

optic disc pit maculopathy (ODP-M), a condition that consists of retinal fluid accumulation, macular holes, and serous retinal detachments.<sup>11</sup> When this occurs, presenting visual acuity is usually 20/70 or worse, but can range from 20/25 to count fingers.<sup>4,12</sup> There are no known precipitating events that lead to ODP-M which can present in 25%-75% of all cases, and tends to occur in the third or fourth decade of life.<sup>11,13</sup>

Much like the formation of the pits themselves, the origin of the fluid seen in ODP-M is another point of discussion. Several theories have been put forth regarding the source of the sub-retinal fluid (SRF), the first being that it comes from the vitreous. Although direct pathways from the vitreous cavity to the sub-retinal space have not been definitively demonstrated, there have been findings to corroborate this theory. In one experiment, India ink was injected into the vitreous cavity of Collie dogs; this ink was later observed in the sub-retinal space. The presence of the ink within the sub-retinal space suggests that a pathway from the vitreous exists. Another study found mucopolysaccharides from the vitreous within optic disc pits, further supporting the notion of the vitreous being the source of the fluid.<sup>11,12</sup>

A second theory suggests cerebrospinal fluid (CSF) can move through an ODP and accumulate in the sub-retinal space. Pits that are at least 1mm deep should be adjacent to the subarachnoid space, where CSF can be found; Krivoy and others felt that the CSF could leave the subarachnoid space and cause a macula-schisis adjacent to the optic disc pit.<sup>14</sup> This theory is further supported by a case in which droplets of silicone oil migrated from the vitreous cavity to the posterior horn of both ventricles in the brain.<sup>12</sup> Furthermore, recent OCT studies have confirmed that communication exists between the subarachnoid and subretinal space.<sup>8</sup>

It has also been proposed that vessels at the base of the pit may be the source of the SRF. An altered permeability of these vessels could allow for an accumulation of fluid. However, when fluorescein angiography is performed, optic disc pits do not hyperfluoresce, which suggests that the vessels are not leaking and, therefore, are not the source of the subretinal fluid seen in ODP-M.<sup>12</sup>

Regardless of the source of the fluid associated with ODP-M, the series of events going from fluid accumulation to a serous retinal detachment is generally accepted.<sup>11</sup> Lincoff and others first described these events in 1988.<sup>15</sup> They postulated that fluid first enters the retinal space and causes a schisis-like inner retinal layer separation. Then, an outer layer macular hole forms beneath the inner

retina. This leads to an outer layer retinal detachment that may present as retinal pigment epithelial detachment. Finally, this detachment increases to the point that it is indistinguishable from a serous retinal detachment.

Studies utilizing an OCT have supported Lincoff's belief that the layers of the retina separate in OPD-M. A 2013 study by Roy et al. looked at 32 eyes with SRF from an ODP. SRF was seen at either the outer plexiform or outer nuclear layer or both in all cases, and there were no cases of isolated sub-retinal space fluid. From these results, they postulated that fluid first enters the outer retina and then may travel to the sub-retinal space immediately or after entering the inner retina.<sup>16</sup>

There is no universally accepted treatment for OPD-M. Initially, bed rest and observation were recommended, and 25% of OPD-M cases were found to resolve spontaneously. Oral corticosteroids have been used to reduce the amount of sub-retinal fluid, but reaccumulation of fluid was often seen after the steroids were discontinued.<sup>11</sup> Focal laser around the nerve without other therapies has been tried, but this also saw a high rate of recurrence.<sup>17</sup> In 1991 Bonnet published a paper that reviewed 25 eyes with OPD-M in which he suggested that vitreal traction played a role in creating the macular separation. He also noted that a posterior vitreal detachment could lead to complete resolution of the retinal detachment.<sup>18</sup> Since then, treatment has centered around inducing a posterior vitreal detachment and then performing a pars plana vitrectomy.<sup>19</sup> A 2013 article in *Retina* described three cases of OPD-M that were successfully treated with a vitrectomy, induced vitreal detachments, and active subretinal fluid drainage through the pit. These pits were then sealed with a scleral flap to prevent repeat accumulation of sub-retinal fluid.<sup>20</sup>

Surgical intervention, especially those performed shortly after the onset of symptoms, has been shown to produce better visual outcomes than observation.<sup>21</sup> The continuous presence of fluid in the macula may lead to degenerative cystic changes that limit the potential improvement of visual acuity.<sup>12</sup> Avci et al. operated on six patients within a month after symptoms were reported and achieved 20/40 vision or better, which suggests that a rapid intervention is preferable.<sup>21</sup>

## CONCLUSION

Congenital optic disc pits are not a common finding. However, clinicians need to be aware of their associated risks and complications. Patients may be completely asymptomatic, or they may have noticeable visual field defects, and a sudden decrease in vision if maculopathy ensues. The treatment for optic disc pit maculopathy is evolving as surgical and technological advances are made.

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