

Diagnosis and Management of Optic Disc Pits

First described in the late 19th century by Wiethe, optic disc pits (ODPs) are anomalous cavitations of the optic nerve.¹ ODPs are rare, and they can be congenital or acquired. Although cases of bilateral ODPs have been reported, ODPs typically present unilaterally. ODPs tend to be solitary, but two or three pits occurring together have also been described.¹ The main complication of ODPs is optic disc pit maculopathy (ODP-M), which can lead to severely decreased visual acuity (VA). The pathogenesis of ODPs is not fully understood, and there is no consensus regarding their treatment.²

Epidemiology

The prevalence of ODP is approximately 1:11,000.² The majority of cases are thought to be congenital (CODPs); however, acquired ODPs (AODPs) may occur secondary to glaucoma or myopia.³ AODPs occur twice as frequently in women and tend to be inferior in location, whereas CODPs typically involve the temporal region of the optic disc.⁴ Although ODPs are most often unilateral, they are bilateral in approximately 15% of cases overall; however, 21% to 48% of AOPD cases are bilateral.¹

ODP-M occurs in approximately 25% to 75% of ODP patients.⁵ This complication manifests as serous retinal detachment, cystic changes, or degenerative pigment changes of the macula.

Etiology and Risk Factors

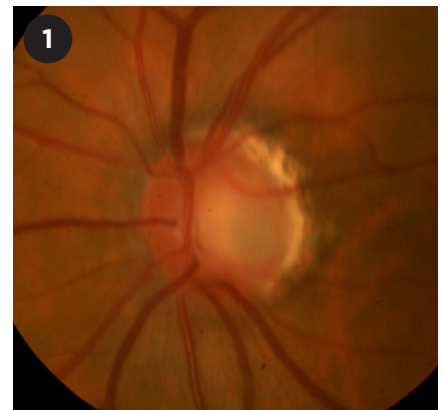
There is no consensus on the embryologic origins of CODPs. Classically, ODPs were thought to represent a more benign variant of optic disc coloboma. ODPs are thought to develop from anomalies in the neuroectodermal folds of the primitive papillae, leading to an abnormal communication between the pit and the subarachnoid space.¹ However, later studies have posited that ODPs are not true colobomas because they are almost exclusively unilateral, sporadic, and rarely inferonasal in location. Moreover, they are typically not associated with iris or retinochoroidal colobomas and usually are not located near the optic fissure.²

Certain rare diseases are associated with an increased risk of ODP and other malformations of the optic disc. They include basal encephalocele, Aicardi syndrome, Alagille syndrome, bilateral renal hypoplasia, and midline neurodevelopmental defects.¹

Pathophysiology

Histologically, an ODP appears as a herniation of dysplastic retinal tissue through a defect in the lamina cribrosa, extending posteriorly to the subarachnoid space. This defect may lead to intraretinal and subretinal fluid in the macula,⁴ although the source of fluid and the mechanism of fluid migration are not fully understood.²

Two commonly accepted fluid

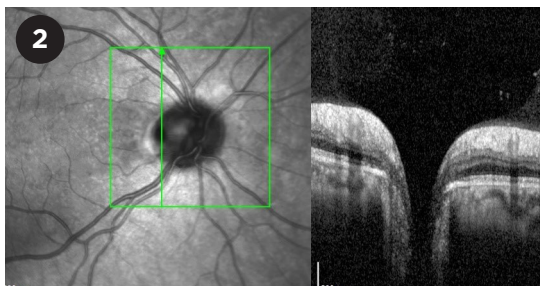


FUNDUS PHOTO. A temporally located gray ODP is seen in a 56-year-old man with primary open-angle glaucoma.

sources are vitreous humor and cerebrospinal fluid (CSF). A less likely source is leakage from vessels at the optic pit base.² Hypothesized mechanisms of fluid migration in ODP-M include vitreous traction and movement of fluid down pressure gradients due to an ODP.² Progressive vitreous liquefaction usually occurs in the third or fourth decade of life, which coincides with typical presentation of ODP-M.

Additionally, pars plana vitrectomy (PPV) has been demonstrated to be a viable therapy for some cases of ODP-M. This suggests that reduction of vitreous traction may play a role in the treatment of some manifestations of ODP-M. However, several optical coherence tomography (OCT) studies have failed to demonstrate an association between vitreous traction and ODPs, and macular detachment may recur after PPV; both of these observations suggest that vitreous traction is

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OCT VIEWS. Horizontal and vertical OCT scans show ODP in the right eye of a 38-year-old man.

not the sole pathologic factor leading to macular detachment in ODP-M.²

A normal eye is a closed system with little difference in pressure between its compartments. However, an ODP forms a conduit that may transmit intracranial pressure (ICP) to the eye from the CSF and vice versa. OCT studies have shown glial tissue and vitreous strands projecting into ODPs, which implies that when ICP is low, vitreous and other tissue may be drawn posteriorly into the pit following the pressure gradient.⁴

Clinical Presentation

ODPs are most often asymptomatic and diagnosed incidentally on fundus examination, although they may sometimes cause visual field defects (most commonly arcuate scotomata).² Generally, ODPs cause symptoms only if they are complicated by ODP-M, which classically presents in the third or fourth decades of life as rapid, progressive visual deterioration due to lesions such as cystic degeneration of the macula and serous macular detachment. However, ODP-M can manifest at any age.²

VA is generally reduced to 20/200 or worse in ODP-M. Spontaneous resolution of macular edema and detachment with recovery of VA is thought to occur in only 25% of cases.¹

Diagnostic Approach

Diagnosis of ODP is mainly based on direct fundus examination and OCT.

Fundus findings. On fundus exam, an ODP is visible as a round depression in the optic disc that appears gray, white, yellow, or black and occupies $1/8$ to $1/4$ of the disc (Fig. 1).^{1,5} Most ODPs are located in the inferotemporal segment of the optic disc, 20% are located centrally, and 10% are located

in other regions. ODPs do not obscure the optic disc margin or the physiological optic cup, which differentiates them from optic disc colobomas.¹

CODPs and AODPs are morphologically similar, thus difficult to distinguish on ophthalmoscopic exam. However, CODPs tend to be temporal, whereas AODPs

tend to be inferior in location.

OCT. OCT imaging of an ODP will show a defect in the lamina cribrosa with herniation of nerve tissue into the pit (Fig. 2). If ODP-M is present, OCT will demonstrate both intraretinal and subretinal fluid collections. The pattern specific to ODP-M is the dual morphology of serous retinal detachment with a schisis cavity and a coexisting detachment of the outer layer of the retinal pigment epithelium.²

Fundus autofluorescence (FAF).

FAF will reveal hyperfluorescence in a granular pattern, as well as subretinal precipitates. Also, areas of serous retinal detachment and inner retinal schisis appear hypofluorescent, but they will become bright after successful vitrectomy and retinal reattachment.²

Visual field defects. In patients with ODPs, visual field defects are variable and usually do not correspond with the location of the pit; paracentral arcuate scotomata are the most common type.⁶

Differential diagnosis. Other conditions to consider in the differential include the following:

- Optic nerve hypoplasia, which is an abnormally small optic nerve head.
- Megalopapilla, which presents as an enlarged optic nerve head with an increased cup-to-disc ratio and a horizontally elongated cup.
- Morning glory syndrome, which appears as a funnel-shaped excavation, an enlarged optic nerve head, and an increased number of disc vessels.
- Optic nerve coloboma, which is characterized by an inferior excavation and is often associated with iris and choroidal colobomas.

In contrast to these entities, ODPs present as round depressions in the disc with a normal or large optic nerve size

and may be associated with maculopathy.¹

Management

Macular edema and detachment secondary to ODP-M were originally treated conservatively. However, because observation alone is often associated with poor visual outcomes, a more aggressive surgical approach is appropriate in some cases.

PPV and adjunctive therapies. PPV is the most widely accepted treatment for serous macular detachment associated with ODP-M. Induction of complete posterior vitreous detachment is likely important because it potentially relieves unidentified tractional forces.² Adjuncts to PPV include internal limiting membrane peeling, laser, and gas or silicone tamponade.⁷

Although laser photocoagulation is sometimes used as monotherapy to treat serous macular detachment in ODP-M, laser alone has been shown to have worse outcomes compared with vitrectomy. It is now more commonly used as an adjunct to vitrectomy and/or gas tamponade.⁷

Intravitreal gas injection with perfluoroethane, sulfur hexafluoride, or perfluoropropane is performed to attempt reattachment of the macula in cases of ODP-related detachment. This technique is often used in conjunction with PPV and laser.⁶

Macular buckling. This surgery involves fixation of a sponge implant to the posterior segment of the globe to produce a buckling effect under the macula. Although it is associated with good outcomes in the management of ODP-related macular detachment, it is a technically difficult surgery with a steep learning curve. Thus, it is not utilized as often as vitrectomy.²

Other techniques. Other approaches have produced promising results.

- Autologous platelet injection over the ODP after PPV has been successful in treating a patient with persistent ODP-related macular detachment.⁸
- Vitrectomy with radial inner retinal partial-thickness fenestration is a newer surgical technique that has been shown to completely resolve subfoveal fluid in 94% of eyes.⁹

- Sealing of ODPs with autologous scleral flaps has been reported to be effective in inducing retinal reattachment and improving VA.²
- PPV and temporal-side single radial optic neurotomy is thought to create a barrier to fluid passage by creating scar tissue and is associated with fluid resolution in 86% of eyes.¹⁰

Conclusion

ODPs are rare cavitations of the optic nerve that may be asymptomatic or may be complicated by ODP-M, leading to significant visual loss. Diagnosis of an ODP is achieved by fundus examination, OCT of the optic nerve, and FAF. ODP-M is managed surgically with PPV, macular buckling, and a variety of other surgical techniques. Surgical management of ODP-M often leads to good visual outcomes. Although ODPs are rare, it is important for ophthalmologists to be aware of this condition and to monitor ODP patients for signs of developing ODP-M.

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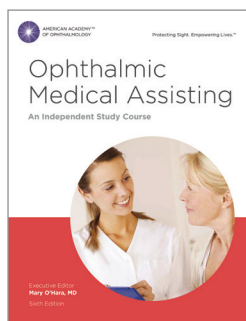
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