

Challenges in Optic Disc Pit Maculopathy Treatment



Many techniques have been described; choose the best one for the individual patient.

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Optic disc pit (ODP) is a rare congenital abnormality of the optic nerve head, first described by Wiethe in 1882.¹ ODP usually appears as a grayish-whitish, round or oval depression of the optic disc (Figure 1A), and it may have different levels of severity. It is a spectrum of congenital cavitory anomalies of the optic disc, ranging from the more common optic pit to the so-called morning glory syndrome (Figure 1B) to coloboma not only of the optic nerve but also of the choroid (Figure 1C). It is also found in the papillorenal syndrome determined by a mutation of the PAX2 gene.

ODP is bilateral in 15% of cases (Figure 2) and occurs equally in men and women with an estimated incidence of 1 to 2 per 10,000 people.²⁻⁵ The most frequent location is in the temporal segment of the disc, but ODP has also been described on the nasal side.^{3,6}

Histopathologically, ODP appears as a herniation of dysplastic retinal tissue into a collagen-rich excavation that can extend into the subarachnoid space through a defect in the lamina cribrosa (Figure 3).⁷ Studies of ODP using OCT have revealed connections between the subretinal and intraretinal space, the perineural space, and the vitreous cavity (Figure 4).⁸

ODP MACULOPATHY

ODP is usually asymptomatic, but a percentage of affected patients (25%-75% depending on the report) may develop a macular serous detachment or a retinoschisis-like maculopathy due to the presence, respectively, of subretinal or intraretinal fluid.^{3,6,9} This condition, known as *ODP maculopathy* (ODPM), causes visual impairment, especially in long-standing cases, in which lamellar or full-thickness macular holes and retinal pigment epithelium atrophy may appear.¹⁰⁻¹²

The exact mechanism of the pathophysiology of ODPM and the origin of the fluid remain unclear. It has been proposed that vitreous or cerebrospinal fluid (CSF) may be the origin of the fluid responsible for ODPM.¹³⁻¹⁵ In the first case, it is posited that the vitreous exerts traction on the macula and optic disc, leading to negative pressure and the subsequent entrance of fluid through the ODP into the submacular space.^{14,15} In the second case, the supposition is that the CSF flows through direct communication between the macular subretinal space and subarachnoid space through the ODP defect.^{10,16,17} This hypothesis was supported by Ohno-Matsui et al,¹⁸ who imaged the subarachnoid space just posterior to the bottom of the ODP using swept-source OCT.

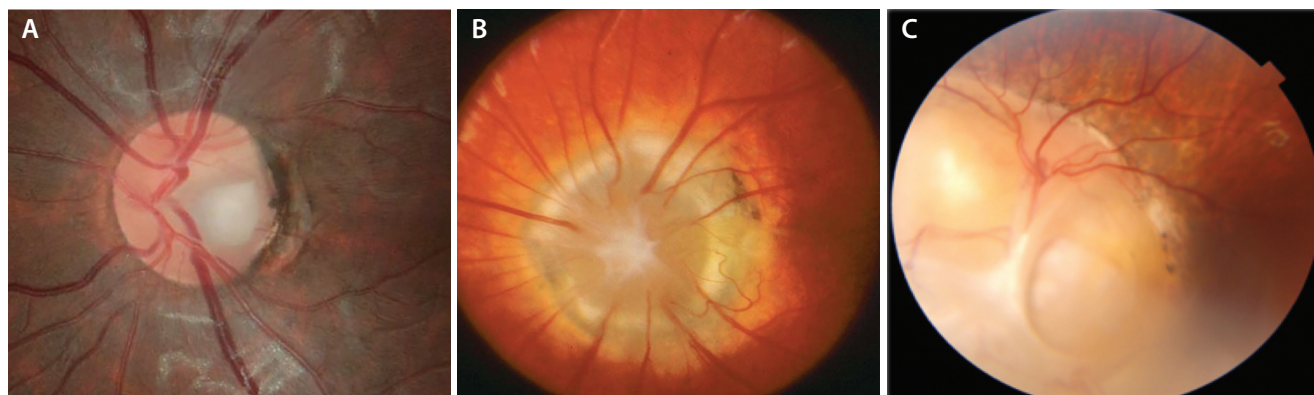


Figure 1. ODP usually appears as a grayish-whitish, round or oval depression of the optic disc (A). ODP ranges from optic pit to so-called morning glory syndrome (B) to coloboma of the optic nerve and choroid (C).

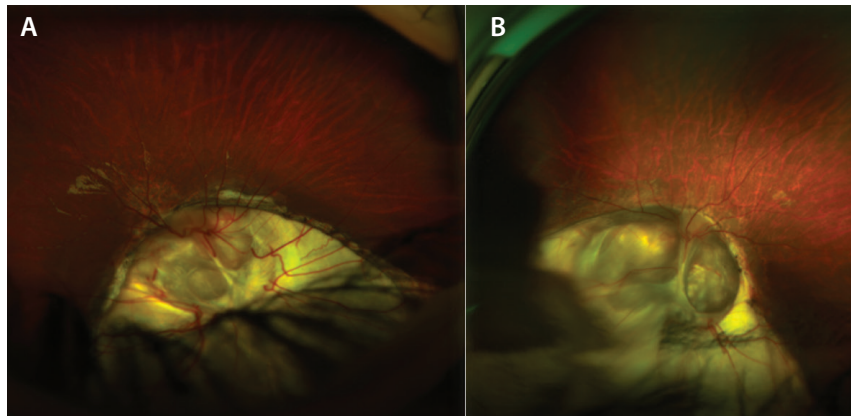


Figure 2. ODP is bilateral in 15% of cases. Retinography of the right (A) and left (B) eye of an 8-year-old boy with bilateral coloboma of the optic nerve and involving a large area of the central inferior and nasal choroid.

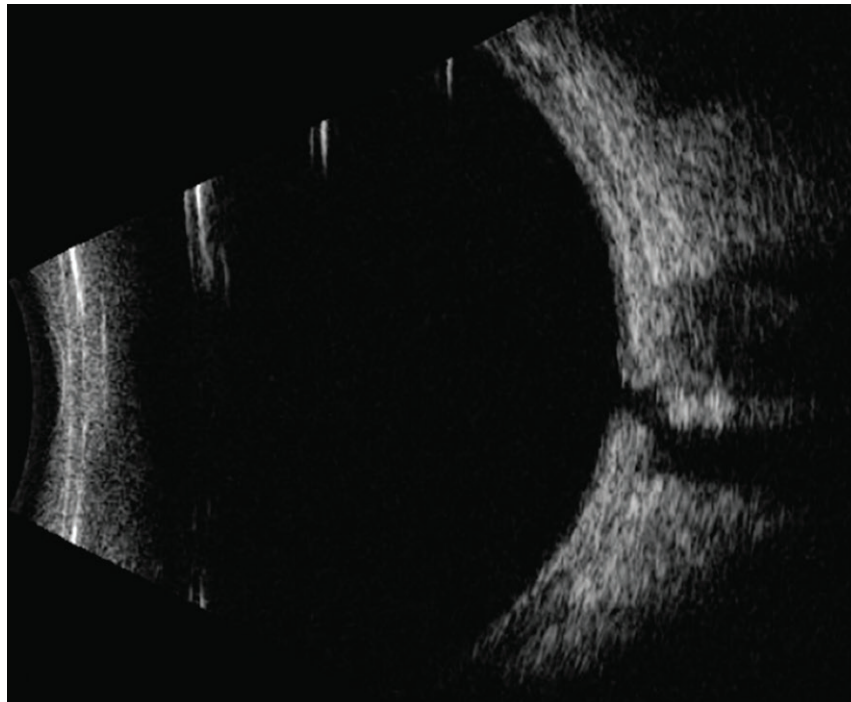


Figure 3. ODP appears as a herniation of dysplastic retinal tissue into a collagen-rich excavation that can extend into the subarachnoid space through a defect in the lamina cribrosa.

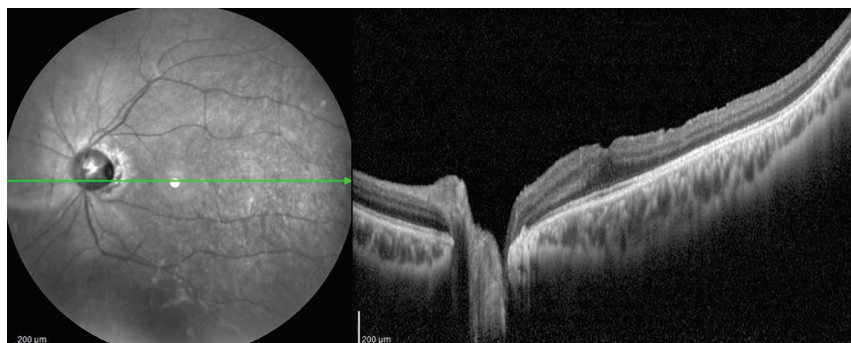


Figure 4. OCT imaging in ODP can reveal connections between the subretinal and intraretinal space, the perineural space, and the vitreous cavity.

RETINA TODAY ON THE ROAD

This article is adapted from a lecture the author presented at the Duke Advanced Vitreous Surgery Course in April. The 2020 meeting will be held April 17-18, 2020, in Durham, North Carolina. Visit MedConfs.com for details.

Moreover, in eyes with ODPM, communication between the vitreous cavity, the subarachnoid space, and the subretinal space was confirmed by the observation of gas and silicone oil migration into the subretinal and intracranial space, respectively, after pars plana vitrectomy (PPV).^{19,20}

In most cases, the fluid follows the pattern described by Lincoff et al,²¹ in which fluid from the ODP first creates a schisis-like separation of the inner retina and then reaches the subretinal space, creating a macular neuroepithelial detachment. Spontaneous reabsorption of fluid is possible in up to 25% of cases according to Gass,²² but relapses are frequent, leading to a progressive deterioration of the macular structures and visual loss. This recurrent trend of ODPM may be explained by the pulsating dynamics of intracranial pressure.²³

TREATMENT OF ODPM

Several treatment alternatives have been proposed for ODPM, including conservative management, laser photocoagulation, macular buckling surgery, gas tamponade, partial thickness retinotomy,²⁴ and removal of glial tissue at the temporal wall of the ODP.²⁵

Another therapeutic approach to ODPM is pneumatic displacement with intravitreal gas tamponade alone or combined with laser.^{21,26,27} Gas tamponade may induce a posterior vitreous detachment, thereby reducing vitreomacular traction, whereas

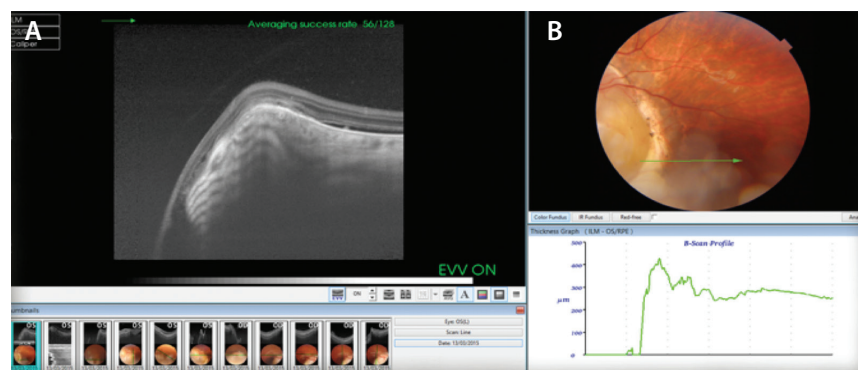


Figure 5. OCT of the left eye of the patient depicted in Figure 2. The horizontal scan of the OCT (A) shows the depth of the coloboma. Identification of the fovea can be challenging in cases of ODP and is not possible in this particular case because it is located inside the coloboma. A mild amount of subretinal fluid is visible. The color photo (B) shows the extensive coloboma and the level of the OCT scan in the extramacular temporal areas shown below.

laser photocoagulation contributes to sealing of the pit.²⁸ This simple and minimally invasive technique has been reported to have a good success rate, although more than one injection is often necessary.^{26,28}

We have tried this technique in 15 eyes with good initial outcomes. In some of these eyes we have observed a relapse. However, in patients younger than 20 years we still consider it preferable to apply this technique first. It can then be repeated years later in the event of relapse, leaving the option of vitrectomy if this treatment fails.

Today, PPV is often the treatment of choice,^{26,29,30} either alone or combined with gas tamponade and/or laser photocoagulation and with or without internal limiting membrane

(ILM) peeling. Multiple studies have reported high anatomic success rate (50%-95%) and good functional outcomes with PPV, with VA improvement in more than 50% of cases.²⁷⁻⁴²

Inducing a posterior vitreous detachment with PPV can relieve the traction exerted by the vitreous on the macula, facilitating the absorption of the subretinal fluid. The application of laser at the edge of the pit helps to seal it, obstructing the communication between the ODP and the retina. However, the need for laser has not been definitively demonstrated.

If the vitreous cavity is the source of fluid in ODPM, then the role played by PPV as an effective treatment is comprehensible. On the other hand, if the subretinal fluid originates from the subarachnoid space, further

explanation regarding the efficacy of PPV is needed.⁴¹ In these cases, the key step for a successful surgery maybe the sealing of the pit.

Several authors have also described stuffing the ODP with materials, including an ILM flap,⁴²⁻⁴⁴ an ILM flap and fibrin glue,⁴⁵ the fibrin sealant Evicel (Johnson & Johnson),⁴⁶ or autologous fibrin.⁴⁷⁻⁴⁹ Additionally, using an inverted ILM flap technique to cover the optic disc, including the ODP but excluding the foveal area, has shown promising results.^{50,51}

We have applied this technique to five patients in the past 5 years when pneumatic displacement did not resolve the pathology. Reabsorption of subretinal fluid might take a few months, even in cases in which the ILM flap is visible in the pit on post-operative OCT (Figure 5).

Macular buckling is another alternative for treatment of ODPM. Theodossiadis et al first described this technique and reported a success rate of about 85%.^{30,41,52-54} The macular buckle pushes the macular surface toward the vitreous chamber, obstructing the entrance of fluid and alleviating vitreous traction.

Macular buckling is considered by many a difficult technique and for this reason is not widely applied. However, surgeons should have many weapons in their arsenals so that they can choose the best one based on the needs of the individual patient.

AT A GLANCE

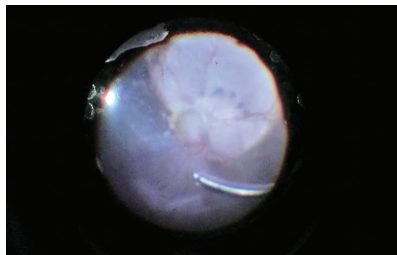
- Optic disc pit (ODP) is a spectrum of congenital cavitary anomalies of the optic disc, ranging from optic pit to morning glory syndrome to coloboma not only of the optic nerve but also of the choroid. Up to 75% of patients with ODP may develop ODP maculopathy.
- ODP maculopathy can cause visual impairment, especially in long-standing cases.
- In a case report, the author describes a challenging repair of an ODP in a young patient.

CASE REPORT

Here, we share the case report of an 8-year-old boy with bilateral coloboma of the optic nerve and the central inferior choroid, as well as the inferior iris as an associated finding (Figures 2 and 5). The patient first developed a macular detachment in the left eye, which quickly developed into a complete retinal detachment caused by fluid coming from the ODP and a suspected macular hole.

This case presented several challenges, including that the macular

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hole was in the area of the coloboma, that the coloboma was very deep, and that there was no retinal pigment epithelium over a large area.

We chose to perform lens-sparing complete PPV (Video). In the literature, similar cases have been managed with silicone oil as a final tamponade. As mentioned above, however, silicone oil can have a track to the CSF (and potentially the brain) in patients with ODP. Therefore, to offer a greater chance of retinal reattachment without the use of silicone oil, we decided to combine PPV with a macular buckle to support the atrophic macular area from the scleral side. This would serve to seal the pit with the buckling effect and to relieve traction and air tamponade.

We still wonder whether performing only the macular buckle and injecting a gas bubble could have solved the detachment. However, the difficulty in determining the cause of the total detachment convinced us to enter the eye. Only during surgery could we confirm that the cause of the patient's pathology was a macular hole into the coloboma.

The surgery was performed in 2012. The retina remains attached, and the boy still retains a BCVA of 20/200 with extrafoveal fixation, the same vision he had before surgery. ■

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