Therapeutic Advances in Ophthalmology

Treatment outcomes of optic disc pit maculopathy over two decades

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Abstract

Aim: To study the long-term outcomes of optic disc pit maculopathy.

Methods: Electronic medical records of 154 patients with optic disc pit were reviewed and 50 patients with optic disc pit maculopathy who met the study criteria were included in the study. Demographic profile of patients, along with clinical characteristics, optical coherence tomography (OCT) features and change in best-corrected visual acuity (BCVA) was recorded. Patients were treated either by observation, barrage laser alone or pars plana vitrectomy (PPV) with optional additional surgical procedures. The primary outcome measures were the change in BCVA and resolution of fluid on OCT.

Results: The mean age of patients was 29.96 years (3–62 years) with a follow-up of 27.16 months. The mean baseline BCVA in observation, laser and vitrectomy group was log MAR 0.94, 0.76 and 0.87 and final BCVA was log MAR 0.9, 0.67 and 0.46, respectively. There was a statistically significant improvement in the final BCVA and reduction in subretinal fluid with resolution of the schisis cavity in vitrectomy group than in other groups. On regression analysis significant association was found between final BCVA with baseline BCVA ($R^2 = 0.815$, $p = 0.002$), use of C3 F8 endotempanode ($p = 0.004$) ILM peeling ($p = 0.012$) and use of triamcinolone (TA; $p = 0.003$). No significant association was found with juxtapapillary endolaser ($p = 0.062$).

Conclusions: In patients with disc pit maculopathy, PPV lead to better functional and anatomical outcomes as compared to laser or observation alone. Use of surgical adjuvants like ILM peeling, TA and C3F8 tamponade improved the outcomes, unlike juxtapapillary endolaser treatment.

Keywords: optic disc pit, pit maculopathy, schisis, treatment outcomes, vitrectomy

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Introduction

Optic disc pit is a rare congenital anomaly attributed to incomplete closure of the foetal fissure. The entity is rare (less than 1 in 10,000 patients seen in the ophthalmic setting) which is bilateral in 10–15% of cases. About 30–75% of cases have a concurrent serous detachment of the macula. Optical coherence tomography (OCT) has contributed in early diagnosis, treatment and better understanding of the disease entity.

Various theories have been proposed for the development of serous macular detachment. Lincoff and colleagues proposed that fluid from optic disc pit creates a schisis like inner layer separation of the retina and the detachment at the outer layer of the macula is a secondary phenomenon. The origin of the fluid requires a more concise understanding as the authors believe either the fluid leaks from the vessels inside the pit or originates from the cerebrospinal region. Gass speculated vitreous traction as a cause of macular detachment. It was believed that the tangential traction at the pit caused passive migration of the fluid into the submacular space leading to optic disc pit maculopathy (ODPM). Spontaneous resolution of ODPM was also described after completion of posterior vitreous detachment (PVD).
The long-term visual prognosis in patients with optic nerve pit and untreated ODPM is generally poor. The reduced visual acuity usually occurs within 6 months of the onset of the serous macular detachment. Juxtapapillary barrage laser in an attempt to stop the fluid causing ODPM has been reported with limited success. Role of pars plana vitrectomy (PPV) with various surgical adjuvants like endolaser and gas injection have been published. Some of the recent authors have used fibrin glue, internal limiting membrane (ILM) grafts and scleral grafts for plugging the pit with variable results.

Despite of multiple prevailing modalities for the management of ODPM, the appropriate timing of intervention and the preferred treatment still remains controversial. This study was undertaken to compare the outcomes of ODPM treated with observation alone, laser photocoagulation or vitrectomy over two decades.

Materials and methods
This study was a retrospective chart review of 154 patients with a diagnosis of optic disc pit who attended Retina Vitreous Services, between January 1994 and May 2018. Eighty-two patients had presented with a optic disc pit and neurosensory detachment (NSD) of the macula. Fifty patients with clinical evidence of ODPM with a follow-up of more than 2 years were included in the study. Patients with concurrent macular disease like central serous chorioretinopathy, cystoid macular edema, choroidal neovascular membrane, pathological myopia and insufficient medical records were excluded from the study. ODPM was defined as serous macular detachment or intraretinal fluid (IRF) in a patient with optic disc pit. An institutional review board and ethics committee approval was obtained to conduct the study. Informed written consent was taken from all the patients and the study protocol adhered to the Declaration of Helsinki. An institutional review board LV Prasad Eye Institute Hyderabad and ethics committee approval was obtained to conduct the study (ethics approval # LEC 11-036).

Preoperative data were collected including demographic profile of patients. All patients underwent a comprehensive ophthalmic examination including best corrected visual acuity (BCVA) intraocular pressure (IOP) measurement, slit-lamp examination and indirect ophthalmoscopy. Fundus photography (Zeiss Visupac® FF4 and FF450-plus, Carl Zeiss, Dublin, CA) and OCT using Zeiss Cirrus HD OCT was recorded and images were analysed wherever available. The fluid on the OCT was categorised into subretinal fluid (SRF) or IRF or a combination. The other related pathologies based on OCT scan like outer retinal hole (ORH) and inner retinal hole (IRH) were also documented. The patients in this study were managed by multiple surgeons with different surgical experience.

Clinical outcomes were measured in terms of the functional outcome as a change in BCVA and anatomical outcome as changes noted in the OCT. Increase in the BCVA of two or more lines was taken as an improvement and decrease of more than two lines as worsening. Anatomical success on the OCT was defined as resolution of SRF/IRF and partial success as the incomplete resolution of fluid.

For this study, patients were divided into three groups: Group 1. Observation with no active intervention but a regular follow-up; Group 2. Delimiting Laser on slit-lamp delivery system; and Group 3. PPV and fluid gas exchange (FGE) with optional additional surgical steps. The treatment protocols used in various groups are as described.

Laser treatment
In all, 532 double frequencies Nd:YAG laser were delivered using a contact lens after anaesthetising the cornea with topical proparacaine. Two rows of faint burns at the temporal margin of the optic disc pit were given with an average power of 150 mW and duration of 150 ms. In children who were un-co-operative for slit lamp delivery, laser indirect ophthalmoscopy was used under general anaesthesia.

Surgical technique
Patients were taken up for surgery under peribulbar or general anaesthesia. Core vitrectomy was done using either 20G, 23G or 25G instruments, and PVD was induced using vacuum of the cutter followed by FGE (20% sulphur hexafluoride, SF6 or 14% perfluoro propane, C3F8). Few patients underwent additional procedures like delimiting endolaser photocoagulation and ILM peeling. Surgical adjuvants like preservative-free triamcinolone (TA) and brilliant blue stain were used as indicated. Patients were advised prone
position for 1 week and topical steroid/antibiotic drops were prescribed in tapering dose.

Follow-up examinations were scheduled at 1 week, 6 weeks, every 4 months up to 1 year and twice a year afterwards.

**Statistical analysis**

The normality distribution of data was ascertained using Shapiro–Wilk test. Kruskal–Wallis test and Mann–Whitney U test was used to compare the baseline and final BCVA in three groups. Logistic regression analysis was done for final visual acuity as dependent continuous variable and other factors as independent categorical variables. Further subgroup analysis was done for OCT data. Kaplan–Meier survival analysis was done for resolution of SRF. The null hypothesis was rejected if \( p \) value less than 0.05.

**Results**

On the chart review of 154 patients with optic disc pit, 82 had presented with ODPM. Six out of 154 patients had bilateral optic disc pit. Of the 82 patients with ODPM, 50 patients who met the study criteria were included in the study. The demographic data are as shown in Table 1.

All of the patients presented with complaints of decreased vision with a mean duration of 10.31 months. Majority of the patients (60%) were correctly referred as cases of ODP; however, 12% of the patients were referred as a case of glaucoma due to disc changes; 12% were clinically suspected as central serous chorioretinopathy (CSCR) by the referring ophthalmologist.

The mean baseline BCVAs was log MAR 0.94, 0.77 and 0.92 in observation, laser and vitrectomy groups, respectively. The mean final BCVA was log MAR 1.00 and 0.50 in observation, laser and vitrectomy groups, respectively (Table 2). The difference in mean change in visual acuity was statistically significant among the three groups [analysis of variance (ANOVA) test]. There was a statistically significant improvement in the final BCVA in vitrectomy group (\( p \) value = 0.002) when compared to the observation group. Improvement in BCVA was also noted in the laser group when compared to the vitrectomy group, but the improvement was not statistically significant (\( p \) value = 0.26). The laser group also showed improvement when compared with the observation (\( p \) value = 0.02).

On regression analysis significant association was found between final BCVA and baseline BCVA (\( R^2 = 0.815, p = 0.002 \)), use of C3F8 tamponade (\( p = 0.004 \)), ILM peeling (\( p = 0.012 \)) and use of TA (\( p = 0.003 \)). No significant association was found with juxtapapillary endolaser (\( p = 0.062 \)). In the vitrectomy group, 70% (14/20) showed two or more lines improvement, while it was 10% (1/10) in observation and 50% (10/20) in the laser group.

Difference in OCT findings between the groups is shown in Table 1. The reduction in SRF and resolution of the schisis cavity based on the OCT was prominent in vitrectomy group than in other groups. In the laser group, even though the minimal increase in visual acuity was noted, the schisis cavity and SRF did not resolve. The difference in the resolution of SRF was statistically significant (\( p < 0.001 \), Chi-square test).

Kaplan–Meier survival curve showed 50% anatomical reattachment at 6 months, and 70% reattached after 12 months (Figure 1). The closure of the outer retinal hole was noted before the resolution of schisis cavities.

Two eyes underwent re-surgery 1 month after initial pars plana vitrectomy with good final visual outcomes. One patient developed a macular hole and other had rhegmatogenous retinal detachment.

**Discussion**

The findings of this study are in agreement with previous studies which have reported that 30–75% of the patients with a optic disc pit develop maculopathy. In the current study, 51.95% of the patients presented with optic disc maculopathy. It is important to note that most of the patients were correctly referred as optic disc pit; however, 12% of the patients were initially misdiagnosed and referred as a case of glaucoma and 12% as central serous chorioretinopathy (CSCR). The optic disc changes in a case of optic pit may mimick glaucomatous changes like increased cup disc ratio and notching. The visual field changes may aid in differentiating the two entities. The reported visual field changes in a case of ODP are paracentral scotoma and enlarging blind spot.11 Serial visual
### Table 1. Demographic details.

| Variable                  | Value                                  |
|---------------------------|----------------------------------------|
| Total patients            | 50                                     |
| Age                       | Mean (SD) 29.66 years (13.20)          |
|                           | Range: 3–62 years                     |
| Follow-up                 | Mean (Months) 27.16 months             |
| Sex                       | Male 39 (78%)                          |
|                           | Female 11 (22%)                        |
| Laterality                | RE 27 [54 %]                           |
|                           | LE 23 [46%]                            |
| Symptoms                  | Reduced vision 46 [92%]                |
|                           | Metamorphopsia 3 [6%]                  |
|                           | Scotoma 1 [2%]                         |
| Referral diagnosis        | Optic disc pit 30 [60%]                |
|                           | Glaucoma 6 [12%]                      |
|                           | CSCR 6 [12%]                           |
|                           | Others 8 [16%]                         |
| Duration of symptoms      | Mean(SD) months 10.31 [19.54]         |
| Follow-up                 | Mean 27.18 [29.81] months             |
| Clinical findings         | Optic disc pit location                |
|                           | Temporal 35 [70%]                     |
|                           | Inferotemporal 14 [28%]               |
|                           | Inferior 1 [2%]                       |
| Baseline OCT (N = 33)     | NSD 13 [39.4%]                        |
|                           | SRF 25 [75.8%]                        |
|                           | Communication to pit 7 [21.2%]         |
|                           | Inner retinal schisis 18 [35.3%]      |
|                           | Outer retinal schisis 24 [72.7%]      |
|                           | Outer retinal hole 14 [41.4%]         |
| Treatment                 | Observation 10 [20%]                  |
|                           | Laser 20 [40%]                        |
|                           | PPV 20 [40%]                          |
| Surgical parameters       | Gauge of PPV                           |
|                           | 20 G 7 [35%]                          |
|                           | 23 G 7 [35%]                          |

(continued)
field tests can be used for follow-up of the patient for documenting progression objectively along with visual acuity. Similarly with the onset of maculopathy the clinical picture may mimic disease entities associated with SRF at the macula, like CSCR; however, careful magnified examination of the optic disc along with OCT may help the clinician in differentiating the two entities. It is important for the general ophthalmologist to be aware of the pertinent OCT features and clinical picture and refer when appropriate.2,12,13

The origin of the IRF causing the maculopathy and the exact pathogenesis of ODPM is still debatable, and the hypothesis proposed by Lincoff still holds, that the fluid seeps from the vitreous cavity through the optic disc pit.3 The role of vitreous traction in the progression of macular detachment also seems appropriate with the improvement in reattachment rates and significant reduction of schisis following vitrectomy than after laser or observation; however, the exact pathogenetic mechanism remains poorly understood. The OCT also shows a bilaminar separation of retinal layers which leads to the schisis cavity formation and subsequent development of NSD. Few cases in this series had isolated outer retinal layer schisis without inner layer schisis. However, all cases of inner layer schisis were associated with outer layer schisis. It is similar to the proposition by Lincoff that macular detachment is secondary phenomenon.7 Four cases of NSD were associated with an outer retinal hole. After surgery, the closure of outer retinal hole preceded resolution of IRF, and this can be taken as a marker of success of the procedure. The complete resolution of schisis cavity may take a longer time.14

In this series, we noticed complete resolution of NSD and schisis cavities after vitrectomy but not following laser treatment. It could also explain the role of vitreous in the formation of schisis cavities and subsequent formation of NSD. The abnormal adhesion of vitreous at the optic disc pit might cause traction, which is eliminated following PPV.9

Congenital cavitary anomalies of the optic nerve head associated with serous detachments of the macula also include optic nerve coloboma and morning glory anomaly.15–17 We found associated retinochoroidal coloboma in 6% of our patients.
In the current series, the youngest patient was 3 years old and the maculopathy in this child resolved on follow-up without any intervention. A conservative approach with closer follow-up should be advocated as reported earlier in children.\textsuperscript{18,19}

Optic disc pits are classically small and most commonly located temporally as noted in this series. The fluid from the disc pit first accumulates within the retinal stroma, most predominantly in the outer plexiform layer. With increasing accumulation, the fluid causes a retinoschisis cavity, but with intact vertical bridging retinal elements. Fluid later enters the subretinal space, either through an obvious outer lamellar retinal hole or possibly through minute, invisible breaks in the outer retina.\textsuperscript{15–17}

Most plausible source of fluid responsible for the maculopathy associated with optic disc pit and other cavitary disc anomalies is the vitreous cavity. It is further supported by India ink studies,\textsuperscript{15} intraoperative drainage of SRF\textsuperscript{20–22} and post-operative subretinal migration of gas and silicone oil.\textsuperscript{23}

Studies have analysed various prognostic markers based on the configuration of fluid and concluded that multilayered IRF is associated with poor prognosis.\textsuperscript{24} The characteristic presence of SRF at the initial presentation is also a prognostic indicator for progressive disease.\textsuperscript{25} Future prospective studies may be planned to objectively assess the configuration of fluid pattern on OCT like SRF or IRF along with assessment of disc pit on automated OCT algorithms. This would help the general ophthalmologist to avoid misdiagnosis and successfully manage maculopathy as it is practised in other macular diseases like diabetic macular edema.\textsuperscript{26–29} Future studies could also look into other imaging modalities like fundus autofluorescence and visual field progression.\textsuperscript{11} Our findings support other studies on the pattern of fluid absorption after the surgical intervention. It is reiterated that complete fluid resorption may take a while henceforth sufficient time (at least 12 months) should be allowed to pass before planning a second intervention.\textsuperscript{30}

Persistence of IRF with initial good BCVA might lead to progressive damage to photoreceptors, more so if the patient is lost to follow-up. It was noted that early surgical intervention in case of progressive deterioration of visual acuity with maculopathy improved both surgical and functional outcomes.\textsuperscript{1}

| Observation  | Laser  | PPV  | Remarks |
|-------------|-------|------|---------|
| Baseline BCVA | Mean (95% CI) | 0.94 (0.63–1.25) | 0.77 (0.57–0.96) | 0.92 (0.7–1.14) | $p = 0.442$ ANOVA |
| Final BCVA | Mean (95% CI) | 1.0 (0.75–1.24) | 0.645 (0.45–0.83) | 0.50 (0.33–0.67) | $p = 0.006$ ANOVA |
| Functional recovery (>2 line improved) | Yes | 1 (10%) | 10 (50%) | 14 (70%) | $p = 0.02$ Chi-square (Yates corr) |
| | No | 9 (90%) | 10 (50%) | 6 (30%) | |
| Anatomical recovery (OCT based) | Yes | 1 (25%) | 5 (38.5%) | 1 (6.3%) | $p = 0.0001$ Chi-square (Yates corr) |
| | Partial | 0 | 0 | 15 (93.8%) | |
| | No | 3 (75%) | 8 (61.5%) | 0 | |
| BCVA better than logMAR 0.8 | Yes | 4 (40%) | 14 (70%) | 18 (90%) | $p = 0.04$ Chi-square (Yates corr) |
| | No | 6 (60%) | 6 (30%) | 2 (10%) | |

ANOVA, analysis of variance; BCVA, best-corrected visual acuity; CI, confidence interval; OCT, optical coherence tomography; PPV, pars plana vitrectomy.
ILM peeling and TA-assisted PVD induction ensures total removal of the vitreous gel and further improves final surgical outcome. In the current series use of 14% C3F8 endotamponade was associated with better final BCVA as compared to 20% SF6, though the SF6 group had fewer patients which needs to be validated in larger randomised trials. However, juxtapapillay endolaser was not associated with any additional improvement in final BCVA and it is not recommended. The patients who had undergone ILM peeling had better final visual and anatomical outcome as reported earlier, based on the assumption that it ensures complete PVD induction and relieves tangential traction. Fovea sparing ILM peeling may be a safer option to avoid deroofing of the macular cyst and ensure total removal of traction due to posterior hyaloid.

Moreover, as compared to the previous decade, use of small gauge sutureless vitrectomy systems and improved visualisation has made the procedure comparatively safe. It takes time to change the established practices; however, good documentation and follow-up of cases regarding the outcome of a particular modality eases the transition. Our findings are in agreement to Avci and colleagues and Bloch and colleagues in supporting the evidence that PPV and gas tamponade led to faster SRF resolution and improvement in vision. Over the last decade, our practice pattern includes offering PPV with C3F8 endotamponade to the patient with documented worsening maculopathy. Juxtapapillary endolaser has not shown any additional benefit and is no longer practised.

Macular buckle is an alternative surgical modality which helps by restricting the fluid ingress into the macula; however, it has a steep learning curve and is not widely practised. None of the patients in this series underwent macular buckle.

Babu and colleagues recently presented findings of a modified procedure using a scleral plug to close the optic disc pit. The positive findings in the group are well understood. However, the group which underwent PPV only in this study did not show improvement as expected.

A recent study on preoperative use of Ocriplasmin to facilitate PVD induction, reported favourable results and reduced complications like retinal breaks.

The retrospective nature of the presented study is the major limitation; however, due to the rarity of the disease, this study over a period of more than two decades describes the broader trend of treatment outcomes. Last two decades also witnessed significant changes in the techniques and instrumentation of vitreoretinal surgery, and the final treatment outcomes contain their effect. Owing to the nonrandomised nature of the study, the possibility of case selection of mild disease in observation and laser group and severe form in the PPV group cannot be ruled out. However, despite this possibility, the outcomes in the PPV group are better. This study over two decades with an extended follow-up period provides us with the useful data on the natural history of the disease. The study was conducted in a mixed cohort of patients of ODPM at tertiary-care apex institute managed by multiple surgeons with different surgical experience. However, these outcomes could be representative of real-world scenario.

Based on the findings of this study, it is recommended that in patients with optic disc pit maculopathy with documented progressive worsening of visual acuity, early PPV with ILM peeling and C3F8 endotamponade results in better anatomical and functional outcomes.

Conflict of interest statement
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