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Case Report

Optic Disc Pit Maculopathy in a Nine -Year-Old Child: A case report

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ABSTRACT

Background: Optic disc pit is a rare optic disc abnormality. It appears as a round or oval, gray, white, or yellowish depression in the optic disc. This disorder can be associated with serous retinal detachment which results on a visual deterioration. Optic pit-associated maculopathy generally occurs in the third and fourth decade of life. The development of optic disc pit maculopathy (ODP-M) in childhood is rare and there are not enough studies on the treatment methods. Therefore, our case report may be helpful in the management of similar cases of paediatric optic disc maculopathy.

Case report: There is a little current consensus on treatment of pediatric optic disc pit-associated maculopathy. In this article, we report a case of an optic disc pit maculopathy in a 9 years old patient, who presented for a routine change of glasses, with a history of gradual deterioration of vision in left eye since 4 months. The fundus examination of the left eye showed a macular retinal serous detachment and a temporal oval optic disc pit gray in color, while ocular examination of the right eye was completely normal. Spectral Domain- Optical coherence tomography (SD-OCT) of the left eye showed a large serous retinal detachment with macular retinoschisis and cystoid macular edema. Spontaneous regression can occur in the pediatric optic disc pit maculopathy after several months.

Conclusion: ODP-M is a rare condition that presents usually in the third or fourth decade of life. In the present case, it appeared in an earlier age than mostly reported. With regular follow up, spontaneous regression eventually happened after several months.

Keywords: Optic Pit; Maculopathy; Case Report; Regression

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INTRODUCTION

Optic disc pit represents herniation of dysplastic retina into a collagen-lined pocket extending posteriorly, often into the subarachnoid space, through a defect in the lamina cribrosa. Most of the reported cases are congenital [(1)].

Acquired optic pits have been documented in normal tension glaucoma [(2)]. Its prevalence is 1 in 11,000 with no gender predilection [(3)].

Although optic pits are typically unilateral, bilateral pits are identified in 15% of cases. In unilateral cases, the involved disc is slightly larger than the normal disc [(4)].

Optic pits commonly involve the infero-temporal quadrant or central portion of the optic nerve head. It can lead to an optic disc pit maculopathy. The pathogenesis of optic pits is unclear. Some authors believe that they represent the mildest variant in the spectrum of optic disc colobomas [(5)].

Twenty-five percent of (ODP-Ms) resolve spontaneously, but the final outcomes of these cases are shown to be poor. Therefore, different treatment modalities such as vitreo-retinal surgery or laser photocoagulation may be preferable over conservative management [(6)].

In this article, we report a case of an optic disc pit maculopathy in a 9 years old Saudi patient presented to the ophthalmology department in New Najran General Hospital. The development of optic pit maculopathy in childhood is rare and there are no enough studies on the treatment methods. Therefore, our case report may be helpful in the management of similar cases of paediatric optic disc maculopathy.

Case report

A 9 years old girl with no medical history, who presented for a routine change of glasses, with a history of gradual deterioration of vision in left eye since 4 months. Her unaided visual acuity was 0.1 in both eyes, corrected to 1.0 OD and 0.7 OS. Anterior segment examination and IOP were normal in both eyes.

The fundus examination of the left eye showed a macular retinal serous detachment and a temporal oval optic disc pit gray in color (Figure 1), with a large serous retinal detachment (Figure 2), while ocular examination of the right eye was normal.

Spectral Domain- Optical coherence tomography (SD-OCT) of the left eye (Figure 3) showed a large serous retinal detachment with macular retinoschisis and cystoid macular edema extending to the disc, the central macular thickness was 499 micrometers.

Regular follow up was decided for this patient aiming for spontaneous regression. After 6 weeks, moderate improvement of the maculopathy was noticed, with reduction of the central macular thickness in the OCT (Figure 4).

After 4 months from the first presentation, complete resolution of the serous macular detachment was shown in the OCT image with improvement of best corrected visual acuity to 0.8 (Figure 5).

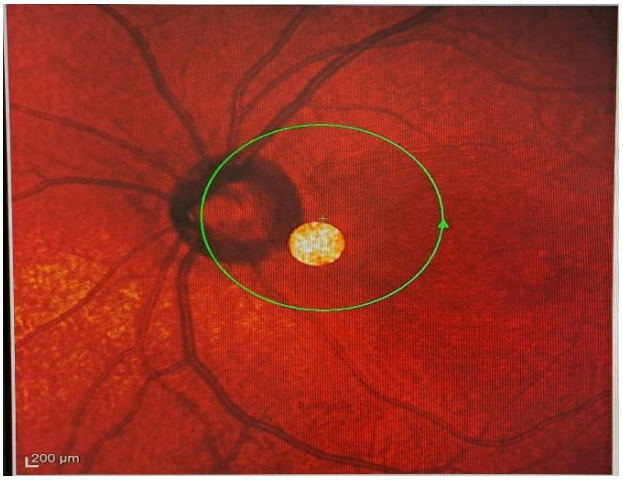

Figure (1): Optic disc pit

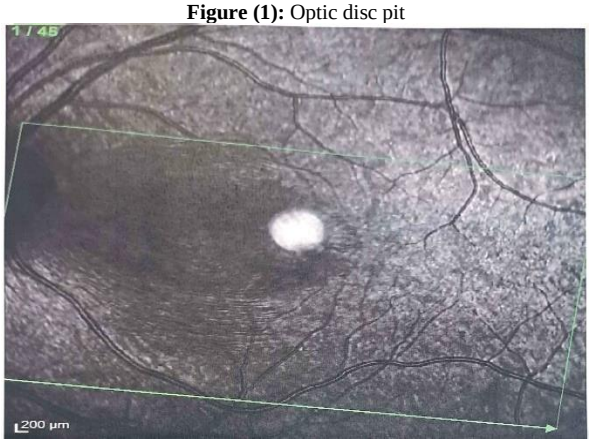

Figure (2): Red free image of serous retinal detachment

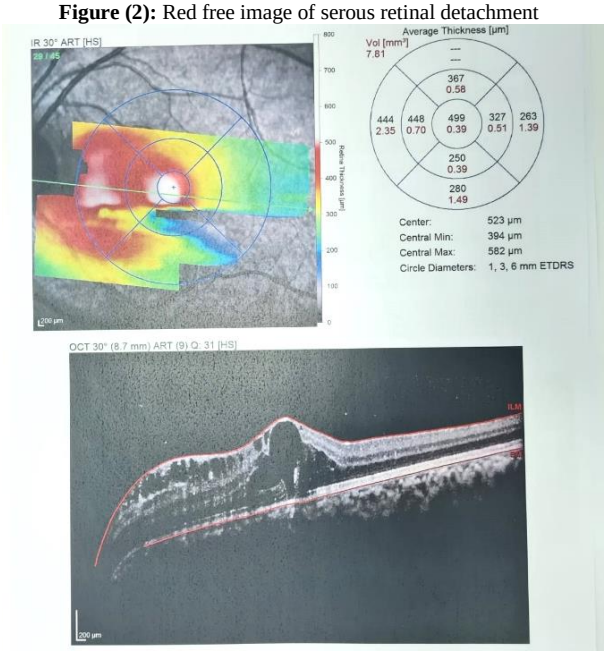

Figure (3): Optical coherence tomography image of the left eye taken at first presentation

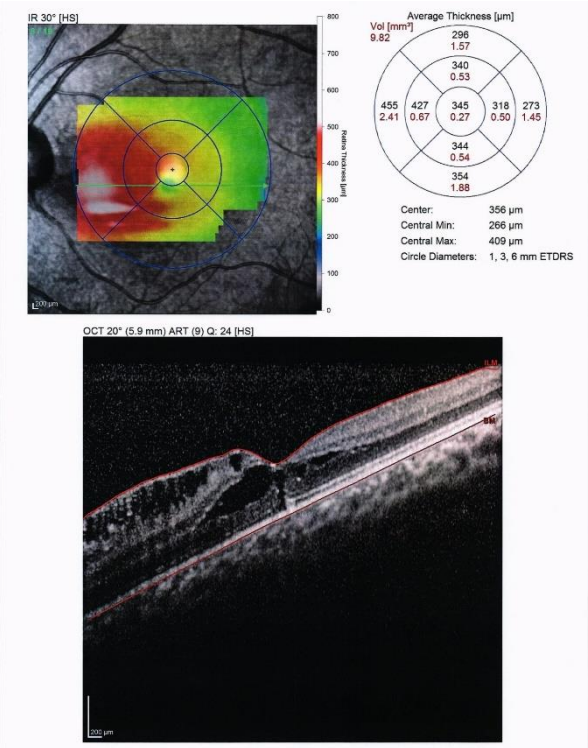

Figure (4): Optical coherence tomography image of the left eye taken after 6 weeks

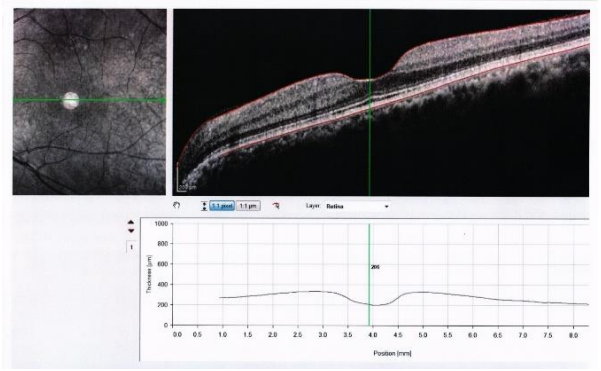

Figure (5): Optical coherence tomography image of the left eye taken after 4 months of the first presentation

RESULTS

The development of optic pit maculopathy in childhood is rare and there are no enough studies on the treatment methods. Therefore, our case report may be helpful in the management of similar cases of paediatric optic disc maculopathy. Optic disc pit maculopathy was first described by **Wiethe in 1882** [7]**.** ODPs are hypopigmented, yellowish, gray-white, oval or round depressions that are usually located unilaterally at the temporal part of the optic disc [8, 9]. ODPs are usually asymptomatic and noticed during routine eye examinations. However, some patients with ODP demonstrate significant macular changes, resulting in irreversible central visual field defects and reduced central visual acuity. These macular changes, including serous macular detachment, cystic degeneration, and degenerative pigment changes, are defined as ODP-induced maculopathy.

Approximately 45% of eyes with congenital optic pits develop serous macular elevations. Until recently, these elevations were thought to represent serous retinal detachments. **Lincoff *et al.*** [10] have proposed that careful serial stereoscopic examination of the macula demonstrates the following progression of events.

1. An inner layer retinoschisis cavity initially forms in direct communication with the optic pit.

2. An outer layer macular hole develops beneath the boundaries of the retinoschisis cavity.

3. An outer layer retinal detachment develops around the macular hole (presumably from influx of fluid from the retinoschisis cavity). This outer layer detachment can be mistaken for pigment epithelial detachment, but it does not hyperfluoresce on fluorescein angiography.

4. The outer layer detachment eventually enlarges and obliterates the retinoschisis cavity. At this stage, it becomes clinically indistinguishable from a primary serous macular detachment.

This abnormality may manifest itself as macular schisis and/or serous macular detachment causing visual deterioration. The percentage of patients with optic disc pit progressing to serous macular detachment is estimated to be around 25–75% [8]. It typically appears during the third or fourth decade, However It appeared earlier in our patient. Pathophysiology of this maculopathy remains unknown and the source of fluid is unclear. The source of fluid in optic disc pit maculopathy may be vitreous or cerebrospinal fluid (CSF) [11]. OCT in patients with this condition showed a micro-communication between the schitic cavity/subretinal space and the optic disc pit [12]. Furthermore, according to a study of **Turkcuoglu and Taskapan** [13]**,** the composition of subretinal fluid was comparable to that of CSF. It has also been proposed that the vitreous fluid is the source of the subretinal/intraretinal fluid in this anomaly. The migration of various dyes between the optic disc pits and the vitreous cavities provides an evidence of a link between the vitreous and the subretinal space [14]. Moreover, in patients with optic disc pits, intravitreal silicone oil and intraocular gas used in vitreo-retinal surgeries have been found in subretinal spaces. Fluid in optic disc pit-maculopathy may also derive from the choroid, via Bruch membrane and peri-papillary atrophy [5]. The role of vitreous traction in optic disc pit maculopathy is controversial. Some authors believe that vitreous macular traction plays an important role in the pathogenesis of optic disc pit Maculopathy [15]. The argument for the role of vitreous traction is that by releasing the vitreous traction with various surgical methods, including vitrectomy with posterior vitreous detachment induction or macular buckling, the maculopathy usually resolves [16].

The symptoms of this condition are non-specific and patients may be asymptomatic. Patients typically present with blurred vision, micropsia and metamorphopsia after developing maculopathy [17]. OCT is the principal test for diagnosis for this anomaly [18]. On OCT, macular schisis and serous macular detachment are typically seen extending from the pit to macular area. Fluorescein angiography has a restricted role in optic disc pit associated maculopathy. In the event of maculopathy due to optic

pit, there is absence of leakage of intravascular fluid into the subretinal space or schitic cavity in fluorescein angiography [19].

The treatment of this condition is challenging, and there is no consensus [6]. Initially, Conservative treatment was recommended, such as oral corticosteroids. However, this approach is no longer a legitimate therapeutic option. Some cases of spontaneous resolution of optic disc-associated maculopathy have been reported, but this rarely occurs and it has also been observed that the final visual outcomes in these eyes on average were worse [20].

Several treatment options have been reported, including laser photocoagulation, Pars Plana vitrectomy [20], pneumatic tamponade with or without laser photocoagulation, inner limiting membrane (ILM) peeling, Macular Buckling, inner retinal fenestration and autologous fibrin and glial tissue removal have been reported.

Conclusions: Optic disc pit maculopathy is a rare condition that presents a challenge in terms of treatment, which usually appears in the third or fourth decade of life. In the management of ODP-M cases, it must be kept in mind that a large serous retinal detachment with macular retinoschisis and cystoid macular edema extending to the disc is possible, especially in pediatric cases such as ours.

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