Case Report

Ocular imaging findings of bilateral optic disc pit in a child

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Abstract

Background: To report a rare condition of bilateral optic disc pit in a child. Case description: A ten-year-old female was admitted with a complaint of headache. Visual acuity was 20/20 in both eyes (OU). Anterior segment examination was normal in OU. Fundus examination revealed optic disc pit (ODP) located temporally with a diameter of 1/5 disc diameter in OU. Intraocular pressure was within normal limits in both eyes. Macular optical coherence tomography (OCT) showed a loss of retinal tissue at the site corresponding to the ODP in both eyes. Retinal nerve fiber OCT revealed decreased RNFL thickness at the temporal side of the optic nerve, corresponding to the ODP in both eyes. The patient and patient’s parents were informed about the disease and called for follow-up examinations every 6 months. In addition, the family was informed about optic pit maculopathy (OPM) and, they were told to return immediately if the patient ever complained of decreased vision in either of her eyes. After a follow-up period of 12 months, visual acuity remained stable, and no complications secondary to ODP were detected. Conclusion: Optic disc pit is diagnosed incidentally unless it is complicated with OPM. The retinal nerve fiber layer thickness is decreased at the side of the optic nerve corresponding to the ODP.

Keywords: Macula lutea, optic disk, optical coherence tomography, retinal nerve fiber layer, retinal detachment.

Introduction

Optic disc pits (ODPs) were first described by Wiethe in 1881 (Georgalas et al, 2011). They are ascribed to incomplete fetal closure of the optic nerve (Oltulu et al, 2011). They usually occur during the first trimester of embryogenesis (Oltulu et al, 2011; Goktas et al, 2010) and are thought to result from a disturbance in the development of the primitive epithelial papilla. In addition, a lack or loss of retinal nerve fibers has been observed at the site of the ODP (Oltulu et al, 2011; Goktas et al, 2010). An ODP is a round or oval-shaped, white, yellowish, grey crater-like depression in the optic disc. The prevalence of ODP is less than 1/11000 patients. Nearly 70% of ODPs are detected on the temporal side of the OD, 20% are located centrally, and the remaining 10% are situated inferiorly, superiorly and nasally (Oltulu et al, 2011; Goktas et al, 2010). The size of the pit varies from 0.1 to 0.7 disc diameter (Georgalas et al, 2011). ODPs may be associated with other abnormalities, such as optic disc coloboma and optic disc enlargement (Goktas et al, 2010; Georgalas et al, 2011). One or two cilioretinal arteries can be seen emerging from the pit base in up to 60% of patients (Oltulu et al, 2011). Unlike optic disc coloboma, an ODP does not affect the margin of the optic disc, and the
physiological optic cup remains distinct (Georgalas et al, 2011). ODP is unilateral in 90% of the patients, and only 10% of patients show bilaterality (Oltulu et al, 2011). In this case report, we present clinical, fundus imaging and optical coherence tomography (OCT) findings of a child patient who was diagnosed as bilateral uncomplicated ODP, which is a very rare condition.

**Case description**

A ten-year-old female was admitted to our outpatient clinic in July 2011 for routine examination. She was in excellent health and had no personal or family ocular or medical history of note. The visual acuity was 20/20 in both the eyes (OU). Cycloplegic retinoscopy findings of the right eye and left eye were both +1.00 diopters. Ocular motility and pupillary responses were normal in OU. Biomicroscopic anterior segment examination was normal in OU. The biomicroscopic fundus examination with a Volk 90 diopter lens revealed a vital optic disc associated with a gray, oval-shaped depression located temporally with a diameter of 1/5 disc diameter in OU (Figure 1-2). Foveal reflex and retinal vessels were normal. Intraocular pressure was 16 mmHg in the right eye and 17 mmHg in the left eye. Macular optical coherence tomography (OCT) showed loss of retinal tissue at the site corresponding to the ODP in both eyes, and no subretinal fluid was detected (Figure 3-4). Retinal nerve fiber layer (RNFL) OCT revealed decreased RNFL thickness at the temporal side of the optic nerve, corresponding to the ODP in both eyes (Figure 5). In light of these findings, the patient was diagnosed as bilateral ODP. Digital retinal photographs were obtained and the patient’s parents were informed about the condition and the patient was called for follow-up examinations every 6 months; however, she was told to return immediately if she ever complained of decreased vision in either of her eyes. After a follow-up period of 12 months, visual acuity remained stable, macular and RNFL OCT findings remained unchanged, and no sign of optic pit maculopathy (OPM) was detected.

**Figure 1:** Optic disc pit is located temporally in the right optic nerve head (White star).

**Figure 2:** Optic disc pit is located temporally in the left optic nerve head (White star).

**Figure 3:** Retinal tissue loss at the site of optic disc pit (red arrows) in macular optical coherence tomography of the right eye.

**Figure 4:** Retinal tissue loss at the site of optic disc pit (red arrows) in macular optical coherence tomography of the left eye.
Figure 5: Retinal nerve fiber layer optical coherence tomography of right and left eyes
Decreased retinal nerve fiber layer thickness at the site of optic disc pit in right (OD) and left eye (OS) marked with red circles and difference between nasal retinal nerve fiber layer and temporal nerve fiber layer thickness marked with blue line and arrow.

Discussion
Optic disc pit is usually asymptomatic in its uncomplicated form, as in this case. Visual acuity is normal in almost all patients, so ODP is detected incidentally in most cases; however, 25 to 75% of patients carry the risk of OPM (Oltulu et al, 2011; Goktas et al, 2010). Until 1988, it was thought that all OPM cases represented serous macular detachment. However, in 1988, Lincoff et al. proposed that, fluid from the pit initially causes an elevation of the nerve fiber layer which leads to a schisis-like inner layer separation, followed by the development of an outer layer macular hole and outer layer retinal detachment (Lincoff et al, 1988). The origin of the subretinal fluid seen in OPM remains controversial. Four different sources have been assumed; vitreous fluid, cerebrospinal fluid, fluid from the leaky blood vessels at the base of the pit and fluid originating from the orbital space surrounding the dura (Lincoff et al, 1988). The onset of OPM is variable. Although ODP is congenital, OPM manifests later in life; the mean diagnosis age for OPM is reported as 30 years (Hirakata et al, 2005). Therefore, in the complicated patients it is assumed that ODP is not a stagnant disease and it progresses in time. A small hole overlying the pit, and the disappearance of the membrane overlying the pit were reported as the potential causes of OPM (Georgalas et al, 2011; Lincoff et al, 1988). Visual acuity loss, blurring, metamorphopsia and hyperopic shift in refraction accompany OPM (Goktas et al, 2010).

No treatment modality has been universally accepted for OPM, since none have been shown to be clearly more effective than the others. The rarity and the challenging nature of the condition have caused this dilemma. Conservative management was used for the initial management, as 25% of the cases resolved spontaneously; however, the visual outcomes were poor in these patients (Georgalas et al, 2011). Several treatment options like bed rest with bilateral patching and oral corticosteroids, laser photocoagulation and/or intravitreal gas injection, macular buckling surgery, pars plana vitrectomy with or without gas injection and internal limiting membrane (ILM) peeling were described (Georgalas et al, 2011). The first regimen was found to be ineffective, but pars plana vitrectomy techniques were found to be very promising (Georgalas et al, 2011). Diab et al., reported successful results from pars plana vitrectomy without ILM peeling in a case of optic pit maculopathy (Diab et al, 2010). Georgalas et al., reported favorable results in patients with optic pit maculopathy who were treated with pars plana vitrectomy and ILM peeling without laser photocoagulation (Georgalas et al, 2011; Georgolas et al, 2009).

Optic disc pit is a congenital defect of the optic nerve head; however, it is usually asymptomatic and is diagnosed incidentally unless it is complicated by OPM. Therefore, ODP may be rarely detected in children. Bilateral ODP is also infrequent. Our case was neither complicated with OPM nor needed a specific treatment. Bilateral ODP is a rare condition, and the macular and RNFL OCT findings of an asymptomatic ODP were clearly documented in this case report.
Conclusion
The optic disc pit can be found in both eyes and it is detected on routine examination in its uncomplicated form. The patients should be carefully informed about the visual symptoms and followed-up closely.

References


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