Case report

Optic disc pit combining with an optic nerve cyst — A case report

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ABSTRACT
Herein, we report a case presenting with the optic disc pit and optic nerve cyst coexistence. A 54-year-old female presented to our office with progressive blurred vision for up to 3 months in her left eye. Upon examination, the best corrected visual acuity in the right eye was 20/20 and in the left eye was 20/100. Fundoscopic examination revealed a black pit at the temporal margin of left optic disc without obvious macular edema. Findings were confirmed later with optical coherence tomography (OCT) examination. Further magnetic resonance imaging (MRI) disclosed a well defined 6 mm x 6mm cystic lesion located at temporal aspect of left retro-bulbar optic nerve within the optic nerve sheath. Compression of left optic nerve was prominent. Visual evoked potential (VEP) test verified dysfunction of left optic nerve. She kept following up for 6 months. Neither maculopathy nor retinal edema occurred during this period. The visual acuity maintained 20/100 in her left eye. The size and location of optic disc pit remained as well. To sum up, in a patient with visual impairment combing with optic disc pit, further imaging surveys should be considered to exclude the possible coexistence of other optic nerve abnormalities.

1. Introduction

According to previous studies, congenital optic disc pit is a rare anomaly occurring in one out of 11,000 people without sex predilection. The appearance is a localized round or oval depression in grey, yellow or black color depending on the amount of glial tissue. The size ranges from 10% to 70% of disc diameter. It is most commonly situated at the temporal aspect of the disc, with about 20% at the central and 10% at other regions, but not outside of it. Bilateral pits are seen in 10–15% of cases and occasionally more than one pit can be found in a disc.1 Though pits usually happen spontaneously in nature, cases suggesting autosomal dominant inheritance have also been reported.2 The congenital optic disc pit is usually asymptomatic unless maculopathy arises, which mostly presents in the third or fourth decade of life.1 However, other causes may also lead to decreased vision. Here, we present a rare case of optic pit coexisting with an optic nerve cyst resulting in compressive optic neuropathy.

2. Case report

A 54-year-old female visited our office for progressive visual impairment lasting for 3 months in her left eye. Upon examination, the best corrected visual acuity was 20/20 in the right eye and 20/100 in the left eye. Intraocular pressure (IOP) was 10 mmHg in the right eye and 12 mmHg in the left eye. Additionally, relative afferent papillary defect was found in her left eye. Nevertheless, she denied any trauma history before this event. No ptosis, proptosis, limited movement of extraocular muscle or any uncomfortable feeling was found. Examination of the anterior segment showed no particular findings. Fundoscopic examination revealed a black pit located at the temporal segment of optic disc (not a glaucomatous disc) in her left eye and no remarkable macular edema (Fig. 1, top left). Findings were later confirmed with optical coherence tomography (OCT) scanning (Fig. 1, top right and bottom left). Due to suspecting retro-bulbar optic nerve abnormalities, ultrasonographic examination was subsequently performed. An optic nerve cystic lesion was surprisingly found (Fig. 1, bottom right). The subsequent visual field test revealed a nasal scotoma in her left eye (Fig. 2, top). Pattern reversal visual evoked potential (VEP) test demonstrated prolonged latencies and reduced amplitudes of N75, P100, N145 components in her left eye (Fig. 2, bottom). Thereafter, MRI demonstrated a well...
defined 6 mm × 6 mm cystic lesion with the same intensity as cerebrospinal fluid (CSF) and located at the temporal aspect of the left retro-bulbar optic nerve within the optic nerve sheath. Moreover, compression of the left optic nerve was prominent (Fig. 3).

Fig. 1. Top left: Color fundus photography showing a black pit located at the temporal margin of optic disc without prominent maculopathy. Top right: optical coherence tomography (OCT) scanning of left eye, initial cross-sectional image of macula revealed no macular edema. Bottom left: OCT scanning of left eye, retinal nerve fiber layer analysis revealed temporal thinner areas outside normal limits and significant loss of ganglion cell complex. Bottom right: Ultrasonographic examination (B-scan) showed a retrobulbar optic nerve cystic lesion adjacent to the optic nerve.

After discussion with the neurosurgeon and radiologist, she refused further intensive management for the optic nerve cyst because of the potential optic nerve damage. In the following 6 months, only IOP-lowering medication was used. During this period, neither

Fig. 2. Top: Visual field test, Full field (central + periphery) examination demonstrated a nasal scotoma in the left eye. Bottom: Visual evoked potentials (VEP) of the left eye illustrated attenuated amplitudes and prolonged latencies of all three parameters.
maculopathy nor retinal edema was found. The visual acuity maintained 20/100 in her left eye. The size and location of her optic disc pit remained as well.

3. Discussion

The pathophysiology of the congenital optic pit is unclear and debatable. For a long time, it has been considered an atypical, mild variant of optic disc coloboma and reported to be associated with other anomalies, such as optic disc, iris and choroid coloboma.3,4 It may result from incomplete closure of the embryonic fissure caused by abnormal development of the primitive epithelial papilla. The congenital optic pit has been reported to be associated with other congenital anomalies including Aicardi syndrome, Alagille syndrome, bilateral renal hypoplasia, basal encephalocele and other midline neurological deformities.1 In addition, optic pit accompanying orbital cyst leading to compressive optic neuropathy has also been reported.5

The most common etiology of visual impairment in patients with optic pit is pit-related maculopathy.1 This is from intra or subretinal fluid accumulation which results in retinal cystic change, retinal pigment change, retinoschisis or serous retinal detachment. The fluid usually extends directly from the pit to macular area. The pit is basically a herniation of dysplastic neuroectodermal tissue into a collagen-walled excavation, extending backward through a lamina cribrosa defect into the subarachnoid space.1 If large enough, reduced retinal nerve fibers and ganglion cells corresponding to the same quadrant may be prominent as well.6 A theory for pit-related serous maculopathy supposes that such abnormality leads to miscommunication between the subarachnoid space and the pit, causing CSF leakage through the pit.1 As for the source of fluid, though still controversial, there are two hypotheses with stronger evidence supporting vitreous fluid originating from the vitreous cavity then penetrating through the pit7 and CSF leakage from the subarachnoid space.8,9 Other theories include leakage from abnormal vessels within the base of pit or from the orbital space surrounding the dura. It is estimated that 25–75% of eyes with pits would develop serous maculopathy. Unfortunately, only about 25% show spontaneous resolution.1

Although the previous treatments were not very promising, they included bed rest with bilateral patching, oral corticosteroids, cryotherapy, diathermy, laser photocoagulation and optic nerve sheath decompression.1,10 In recent years, the trend of management has been changed to a combination of pars plana vitrectomy, posterior vitreous detachment induction and gas tamponade.1 Internal limiting membrane (ILM) peeling can be also considered as well since both vitreous and ILM traction are now thought to be important factors for pit-related maculopathy.1

In our patient, her optic disc pit was located at the temporal margin of disc, the same as the most common region mentioned previously. Nevertheless, an orbital cystic lesion was found accidentally. The similar intensity with CSF suggested developmental abnormality. Furthermore, her visual impairment resulted from compression of optic nerve instead of pit-related maculopathy. These were confirmed by OCT and VEP examinations. The patient...
received only medical treatment and she refused intensive managements after we reviewed the literature and had a discussion with her. Her visual acuity remained stable after 6 months. From the literature review, there are only two cases that mentioned the optic disc pit coexisting with an optic nerve cyst.11,12 The cause of the cyst is unknown but may be the consequence of acquired fluid accumulation from the pit or developmental abnormality. For the latter, as with the microphthalmos and optic disc colobomas being related to some congenital colobomatous cysts, the optic disc pit may also belong to this entity. Due to the rarity, no successful treatment of the pit-related optic nerve cyst has been reported. Possible intensive managements include surgical fenestration and computed tomography (CT)-guided aspiration. Up to now, no patients are willing to receive these risky approaches. As for other potential management strategies including pars plana vitrectomy, laser photocoagulation, IOP-lowering agents and corticosteroid, more cases are required to understand whether these would be beneficial.

In conclusion, for patients with optic disc pit, ophthalmologists need to be wary of the combined optic nerve abnormalities. Further imaging studies may be necessary to complete the evaluation and diagnosis.

References