

OPTICAL COHERENCE TOMOGRAPHY OF A COEXISTING OPTIC NERVE CYST AND OPTIC DISC PIT: A CASE REPORT

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We describe a rare case of an optic pit located over the nasal portion of the disc that was independent of a coexisting optic nerve cyst. A healthy 35-year-old patient was referred to our clinic seeking alternative treatment options because his previous ophthalmologist had suggested evisceration and orbital surgery for a suspected orbital malignancy. Our examinations revealed a gray lesion located within the right disc cup that was associated with a round lesion at the retrobulbar optic nerve. Based on optical coherence tomography and magnetic resonance imaging findings, we believe that our patient exhibited an optic disc pit coexisting with a congenital optic nerve cyst. Because the patient had very good central vision and the lesions were benign, we recommended continued observation for this patient.

Key Words: disc pit, optical coherence tomography, optic nerve cyst
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Congenital optic disc pits, optic disc coloboma and morning glory anomalies can be classified as cavitary anomalies of the disc. The clinical presentation varies from asymptomatic individuals to those who are completely blind, based on the involvement of the nerve fiber layer and macula. The presence of congenital optic disc pits in combination with retinal schisis and subretinal detachment has been demonstrated by high-resolution optical coherence tomography (OCT) [1,2]. Mann suggested that the pits are due to incomplete closure of the upper end of the embryonic fissure or an innate anomaly in the optic vesicle wall [3]. The coexistence of an optic pit and an optic nerve cyst in this case may be incidental or associated with a developmental malformation. The optic nerve pit and optic

nerve cyst may be anatomically connected by the subarachnoid space [4,5]. This report presents diagnostically valuable and complete images in this case.

CASE PRESENTATION

A healthy 35-year-old man attended our ophthalmologic clinic requesting a second opinion because his prior ophthalmologist suggested that he needed eye surgery after orbital imaging had revealed a retrobulbar optic tumor. The patient reported nonspecific complaints such as mild ocular discomfort and dizziness. He did not complain of metamorphopsia or decreased vision. Upon ocular examination, his best corrected visual acuity was 20/25 in the right eye and 20/20 in the left eye. The anterior segment and intraocular pressure in each eye were normal. The patient's pupils were isocoric without a relative afferent pupillary defect. A fundus examination revealed a well-defined, gray-colored lesion on the nasal portion of the optic disc and subtle pigment changes in the macular



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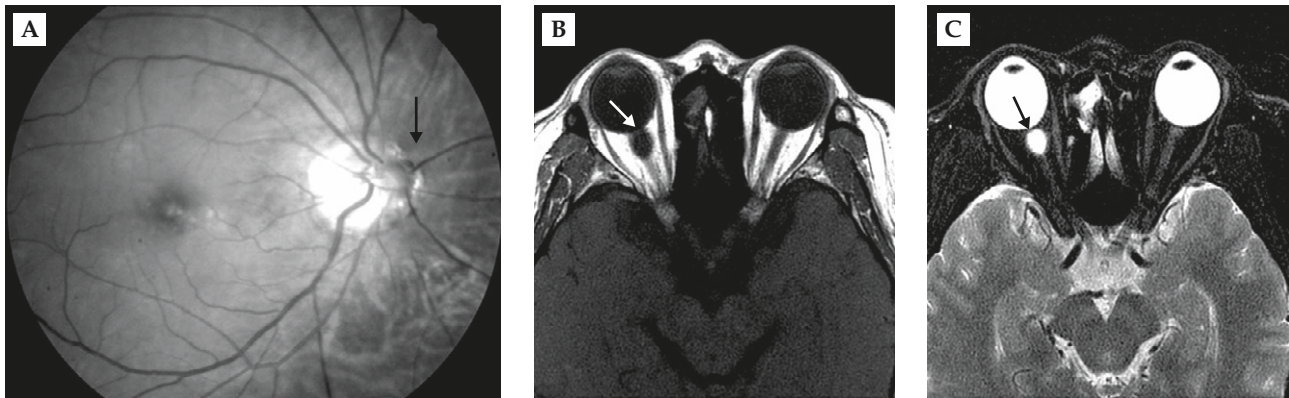


Figure 1. (A) Color fundus photography shows a gray lesion (arrow) on the nasal margin of the disc with subtle macular depigmentation in the right eye. (B and C) The axial T1- and T2-weighted images show a well-circumscribed, non-enhanced retrobulbar cystic lesion of the optic nerve (arrow) that is approximately 6 × 7 mm in size.

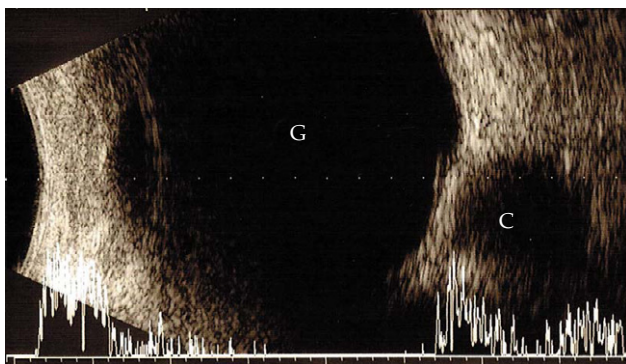


Figure 2. B-mode ultrasonography shows a cystic structure (C) behind the globe (G).

area of the right eye. The pigmentation in the left eye was normal (Figure 1A). The disc lesion was not elevated, showed no neovascularization, and had clear margins without infiltration. In addition, magnetic resonance imaging (MRI) revealed a well circumscribed cystic mass, measuring approximately 6 × 7 mm² in size, located in the intra-orbital part of optic nerve. The cyst had similar signal intensity to that of the vitreous, with a low intensity on T1-weighted images and a high intensity on T2-weighted images without enhancement (Figures 1B and 1C). B-mode ultrasonography showed a cystic structure behind the globe (Figure 2). The communication between the optic pit and the retrobulbar cyst was too small to be detected on the sonogram and MRI. The central visual field examinations revealed a temporal defect with a marked border in the right eye (Figure 3). Fluorescein angiography showed a well-defined patch lesion with hypofluorescence at the nasal margin of the optic disc. OCT studies revealed a deep depression in the area of the

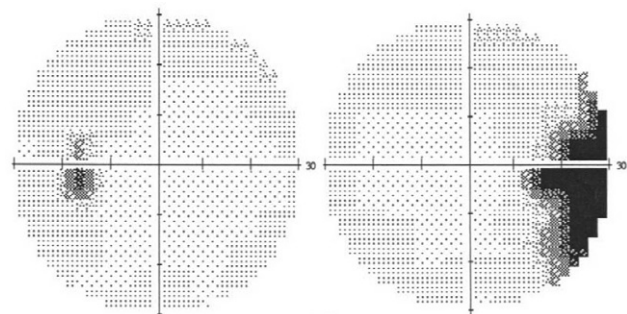


Figure 3. Central 30-2 Humphrey visual field examinations revealed a normal visual field in the left eye and a temporal scotoma in the right eye.

pit, not an elevated lesion. A large defect on the full layer of the retina and choroid over the disc head was also noted. There was no macular detachment (Figure 4). The patient was counseled about the possibility that the cyst may be a benign tumor and continued observation was recommended.

DISCUSSION

A patient with an optic nerve cyst associated with temporally located optic disc pits was reported by Theodossiadis et al [6]. While their paper did not describe disc pit findings on OCT, our results from OCT examinations showed that the unusual nasally located disc pit was a congenital anomaly rather than a malignant lesion.

The incidence of a coexisting optic nerve cyst and optic disc pit is unknown and may be overlooked since ophthalmologists typically do not perform

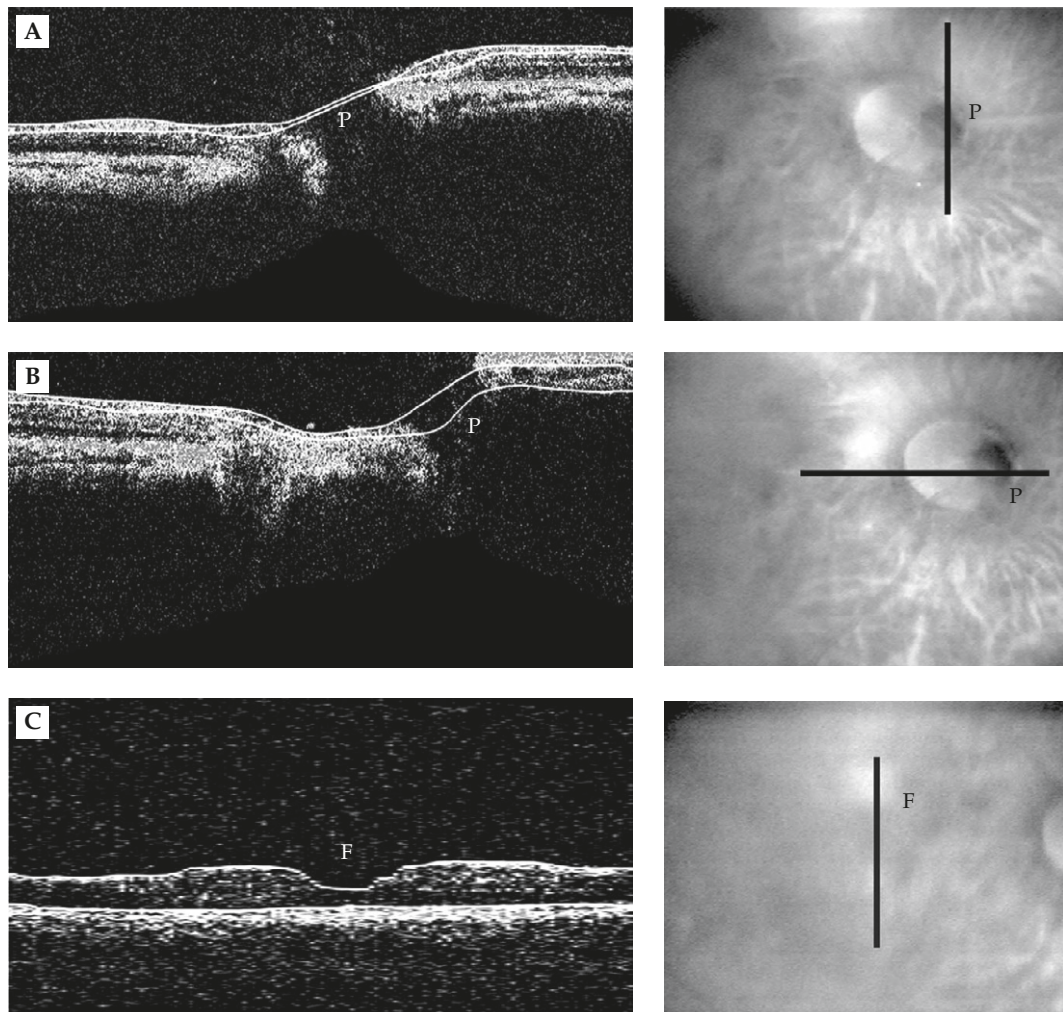


Figure 4. (A and B) The vertical and horizontal linear optical coherence tomography scan through the pit site (P) showed a deep depression in the pit on the nasal aspect of the disc. The pit lesion presented a full layer defect of the retina and choroids. (C) The vertical optical coherence tomography scan through the fovea did not show inner layer schisis of the retina and fovea (F).

neuroimaging studies for patients with optic disc pits [6,7]. B-mode ultrasonography and MRI can be used to confirm the communication between a pit and an optic nerve cyst [6,7]. High-resolution OCT can define the features of the optic disc pits, such as the membrane tissue spanning the cup, which may represent neuroectodermal or astroglial tissue [2]. Moreover, advanced imaging studies, such as OCT and MRI, may help us to detect this rare coexistence.

The locations of the pits vary in individuals, with most pits located in the temporal (72%) and central (21%) regions of the disc. However, only 2% of pits were located in the nasal portion of the disc in a large study of 75 pit cases [1]. Although only 52% of the cases were characterized by pits in association with retinal detachment, in almost 95% of cases with retinal

detachment, the pits were temporally located in the disc [1]. Typically, pits located in the nasal portion of the disc, as in our patient, are less likely to cause visual disturbances.

The fetal fissure starts to close during weeks 6–7 of embryonic development. If this closure process fails, a typical coloboma without an overlying retina develops and a colobomatous cyst subsequently forms [3]. Greear suggested that the pits are atypical colobomatous defects [8]. However, our patient had a simple disc pit associated with a nerve cyst and not a typical coloboma or a colobomatous orbital cyst. We believe that the association in our patient was only a developmental defect, although the occurrence of systemic abnormalities and hereditary ocular disease in conjunction with optic pits have been reported [9,10].

In conclusion, the very rare location of the pit, the nasal portion of the disc head, coupled with a rare optic nerve cyst, may hinder diagnosis. High-resolution OCT and MRI are helpful in reaching diagnosis.

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視神經盤小凹合併良性視神經囊腫之罕見病例

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一位健康無病史的 35 歲男性接受外院眼科檢查發現右眼視神經盤偏鼻側位有一處凹陷性灰黑色病兆及同側眼窩腫瘤，病人曾被告知此病兆為惡性視神經黑色素瘤合併眼窩視神經轉移而被建議眼球摘除併眼窩手術。病人至本院眼科尋求其他治療意見。我們利用高解像度光學同調斷層掃描術及眼窩磁振造影發現此病兆實為極少見的先天性視神經盤發育不良所造成的小凹合併相連性良性視神經囊腫，建議病人不需手術治療且定期追蹤。

關鍵詞：視神經盤小凹，光學同調斷層掃描術，視神經囊腫

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