CASE REPORT

Optic disc pit with coexisting optic nerve cyst: a case report.

Koumpoulis I[1], Gorgoli K[2], Benekos K[2], Kuulasmaa E[3], Ranta K[4], Katsanos A[2]

[1]Ophthalmology clinic, General Hospital of Ioannina "G. Hatzikosta"
[2]Department of Ophthalmology, University Hospital of Ioannina
Ioannina, Greece
[3]Department of Ophthalmology, South-Savo Hospital, Mikkeli
[4]Coronaria Eye Hospital, Helsinki
Finland

Abstract

Purpose: To present the case of a 23-year-old woman with an optic disc pit coexistent with an optic nerve cyst in her left eye and to highlight the importance of optic nerve imaging in this cavitary optic disc disorder.

Case Presentation: The patient was referred with the diagnosis of an optic disc pit in her left eye and a left-sided feeling of tension at the head and neck without any visual deterioration. The best-corrected visual acuity was logMAR 0.0 bilaterally, and intraocular pressure was within normal limits. On fundoscopy, a dark-colored optic pit was present in the temporal segment of the optic disc with respective neuroretinal rim thinning in the left eye. B-mode ultrasonography revealed a cystic lesion behind the globe, while magnetic resonance imaging confirmed the diagnosis of an optic nerve cyst. Since there was no visual deterioration, a decision for regular follow-up was made.

Conclusion: An optic disc pit can be associated with an optic nerve cyst. Our report underscores the importance of optic nerve imaging in comprehensively assessing this cavitary optic disc disorder. HIPPOKRATIA 2024, 28 (4):176-179.

Keywords: Optic disc pit, optic nerve cyst, cavitary optic disc disorder

Corresponding author: Andreas Katsanos, MD, PhD, FEBO, Ophthalmology Department, University of Ioannina, Ioannina, Greece, tel: +302651099706, e-mail: katsanos@uoi.gr

Introduction

Wiethe's 1882 report is the earliest known description of what we now recognize as optic disc pits (ODPs)[1]. Nowadays, OPDs are included in the spectrum of cavitary optic disc anomalies[2] along with morning glory syndrome and optic disc colobomas. ODPs are rare, having an estimated prevalence of 0.02 % with no sex preponderance[3,4]. While typically unilateral, approximately 15 % of cases are bilateral[5]. Most unilateral pits are sporadic, but an autosomal dominant inheritance pattern has also been suggested[6].

Usually, ODPs are asymptomatic and constitute incidental findings during fundoscopy. They are primarily single and located at the inferotemporal segment of the optic disc as a small yellow, greyish, or black cavitation[5]. Occasionally, ODPs can coexist with optic nerve cysts (ONCs), often causing visual deterioration[7]. This report presents an unusual case of a young woman with an ODP and an ONC in her left eye.

Case presentation

A 23-year-old woman was referred to our center with the diagnosis of an ODP in her left eye. No visual deterioration or any other ocular symptoms were reported, but the patient was remarkably stressed due to a left-sided feeling of tension in the head and neck. Her medical history included the excision of a benign breast tumor and two episodes of atrial fibrillation treated with direct-current cardioversion. She had a family history of a brain aneurysm.

On clinical examination, the best-corrected visual acuity was logMAR 0.0 bilaterally. Slit-lamp examination revealed no pathology. The intraocular pressure was 18 mm Hg and 20 mm Hg in the right and left eye, respectively. There was no relative afferent pupillary defect, and ocular motility was normal. Color vision and light brightness perception were unremarkable. On fundoscopy, in the left eye, a dark-colored pit was present in the temporal segment of the optic disc (Figure 1). The macula appeared normal, and no intraretinal or subretinal fluid was detected during fundoscopy or optical coherence tomography (OCT).

Enhanced depth imaging OCT of the left optic disc revealed a depression around the pit (Figure 2), corresponding to the temporal thinning of the retinal nerve fiber layer (RNFL) (Figure 3). No defect could be documented in the visual field of the left eye (Figure 4). B-mode ultrasonography of the left eye revealed a cystic lesion behind the globe and within the optic nerve (Figure 5). Magnetic resonance imaging (MRI) along with MRI

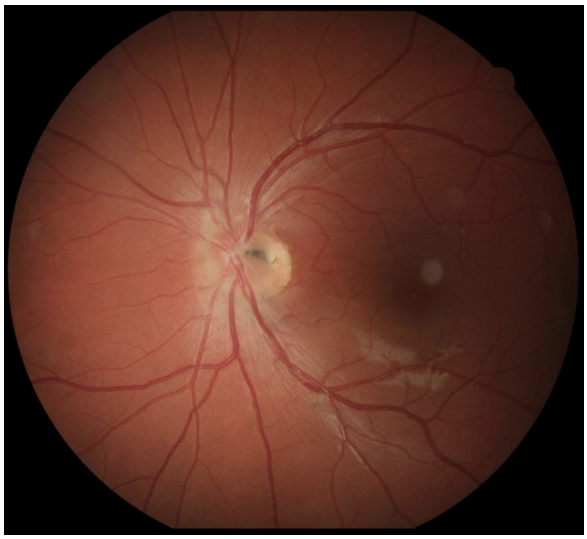

Figure 1: Image of the patient's left eye fundus showing a dark-colored pit at the temporal segment of the optic disc.

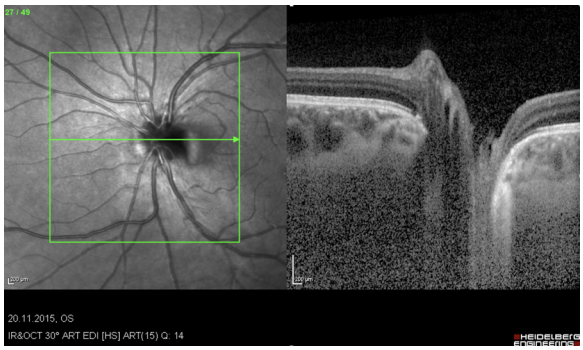

Figure 2: Enhanced depth imaging optical coherence tomography of the patient's left optic disc revealing a depression around the pit.

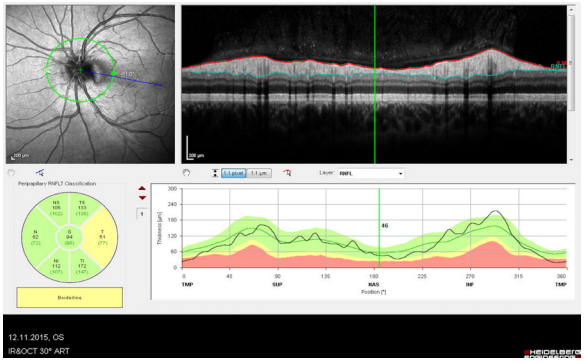

Figure 3: Optical coherence tomography image demonstrating thickness measurement of the retinal nerve fiber layer showing a temporal thinning of the retinal nerve fiber layer.

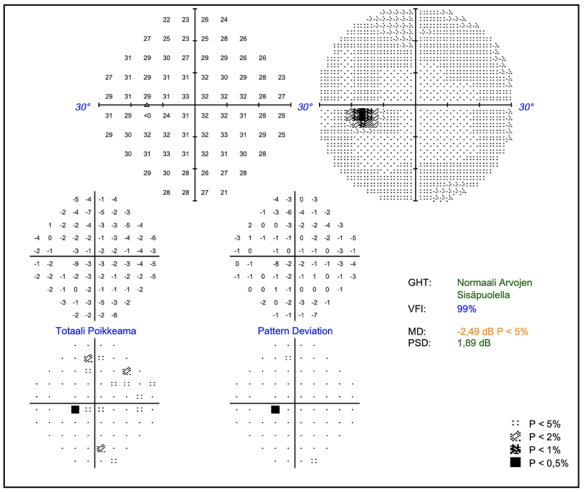

Figure 4: Visual field measurements of the patient's left eye documenting no defects.

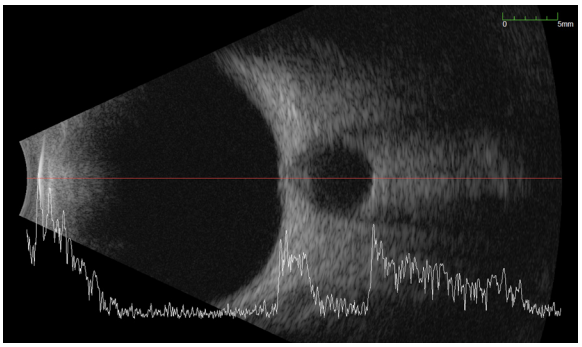

Figure 5: B-mode ultrasound of the patient's left eye, revealing a cystic lesion behind the globe and within the optic nerve.

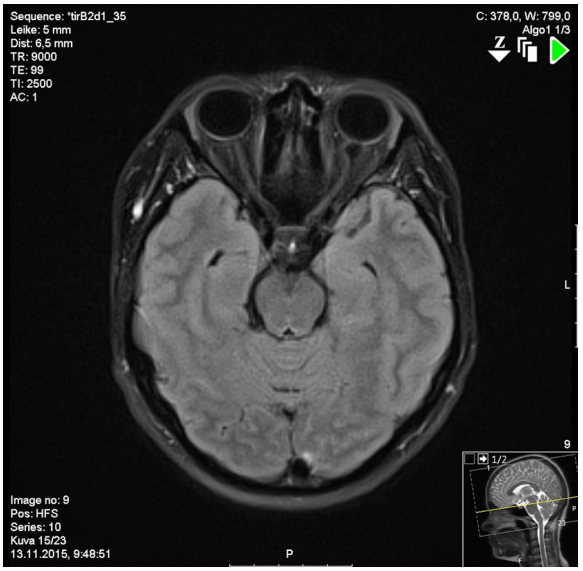

Figure 6: Fluid attenuated inversion recovery (FLAIR) magnetic resonance imaging with fat suppression. A low-intensity cystic lesion can be identified within the sheath of the left optic nerve.

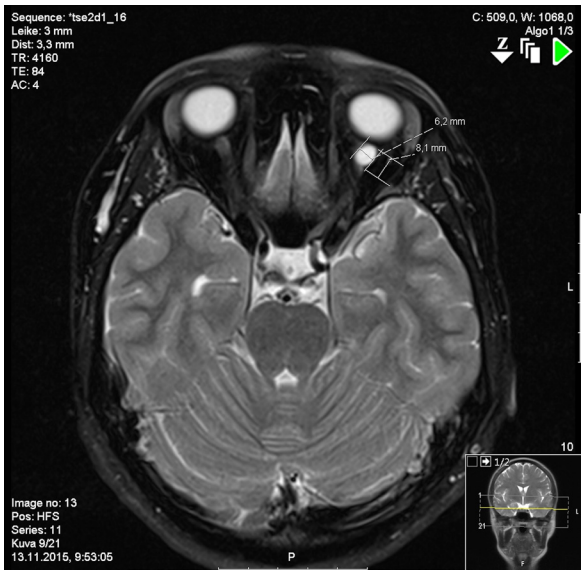

Figure 7: T2-weighted magnetic resonance imaging with fat suppression. The cystic lesion is well-defined and highly intense, measuring 6.2 x 8.1 mm.

angiography were requested. A well-demarcated cystic lesion 6.2 x 8.1 mm in size was revealed within the sheath of the left optic nerve. In fluid attenuated inversion recovery (FLAIR) MRI with fat suppression (Figure 6), the cystic lesion appeared to have low intensity, in contrast to the T2-weighted images with fat suppression (Figure 7), where the intensity of the cyst was high. No lesion enhancement was observed after intravenous administration of a contrast agent. MRI angiography of the brain did not document any intracranial pathology.

The diagnosis of an ODP associated with an ONC was established. Since there was no visual deterioration and both the ODP and cyst were incidental findings, a decision for regular follow-up was made, and no surgical intervention was performed. It was explained to the patient that her symptoms were unlikely to be caused by these findings. The patient reported that her emotional stress was reduced following the work-up and reassuring consultation, and her symptoms subsided over the next few days.

Discussion

The pathogenetic mechanism of ODP remains uncertain. For a long time, ODPs were considered a type of optic disc coloboma resulting from the defective closure of the upper end of the embryonic optic fissure during embryogenesis[5].

Typically, uncomplicated pits are asymptomatic. However, they are sometimes associated with important macular changes such as serous retinal detachment and retinoschisis. This type of maculopathy, known as ODP maculopathy (ODP-M), affects 25 % to 70 % of patients with ODPs[8], most commonly during the third and fourth decade of their lives[5]. The location of the pit plays an important role in the development of ODP-M, as patients with temporal pits are more susceptible to retinal detachment and retinoschisis[5]. Nevertheless, the precise mechanism behind ODP-M remains uncertain, as the origin of the retinal fluid and the mechanism of the retinal detachment have not been clarified yet. The fluid has four proposed origins: the vitreous cavity, cerebrospinal fluid (CSF), leaky capillaries at the pit base, and choroid across Bruch's membrane[9]. Among these, the most widely accepted theory points towards the vitreous cavity. Typical visual field defects related to ODPs are arcuate scotomas, as the pit dislocates the nerve fibers in and around the optic disc[9].

In the reported case, the patient did not suffer from ODP-M, and the visual field of her left eye exhibited no defects. However, the ODP was accompanied by an ONC. The combination of these two clinical entities in the same eye is scarce, and its exact incidence is unknown[10,11]. Similar cases have possibly been left unnoticed as B-mode ultrasonography and MRI are necessary for the diagnosis of orbital cysts, but, typically, clinicians do not perform this kind of neuroimaging studies as a routine in cases of optic disc pits[10]. To the best of our knowledge, only a few similar cases have been reported, and none of them could elucidate the mechanism of the simultaneous presence of both an ONC and an ODP[7,10,11]. Congenital cavitary anomalies of the disc, like pits, may sometimes lead to abnormal connections between intraocular and extraocular spaces. It has been hypothesized that ODPs may communicate with subarachnoid and subretinal space, leading to cyst formation either in the optic nerve or the retina[11,12]. Thus, the cystic fluid may be either liquified vitreous or CSF[13]. Nair et al. aspirated the cystic fluid from a large ONC associated with an ODP in a six-month-old child[13]. However, the biochemical analysis of the aspirated material did not reveal any particular similarity with CSF[13].

ONCs may have different aetiologies: related to meningiomas, post-traumatic or idiopathic[14]. Although they are linked to a wide range of nonspecific orbital or neurological symptoms (e.g., headaches, visual deterioration, proptosis) and findings (e.g., optic disc oedema), they may also be completely asymptomatic. Other conditions included in the differential diagnosis of ONCs are meningiomas, optic nerve gliomas, and vascular malformations such as hemangiomas[14].

The absence of reliable and extensive scientific data for such cases renders counseling and decision-making challenging. Surgical fenestration and computed tomography-guided aspiration might be potential therapeutic choices. Cystic fluid aspiration might have a transient result as the fluid may re-accumulate[13]. Surgical cyst removal could be an option in cases where vision is threatened (i.e., decline in vision, proptosis). However, observation should be advised in asymptomatic or oligosymptomatic patients due to the high risks of such surgical procedures[7].

Our case highlights the importance of optic nerve imaging in eyes with ODP, as this entity can be associated

with an ONC. In such cases, the diagnosis is established using B-scan ultrasonography and MRI of the orbits. Clinicians should remember that the increasingly common use of modern imaging modalities might lead to the increased detection of such lesions.

Conflict of interest

The authors declare no conflicts of interest.

References

1. Wiethe T. Ein Fall von angeborene Differmitat der Sehnervenpapille. Arch Augenh. 1982; 11: 14-19.
2. Jain N, Johnson MW. Pathogenesis and treatment of maculopathy associated with cavitary optic disc anomalies. Am J Ophthalmol. 2014 ; 158: 423-435.
3. Wang Y, Xu L, Jonas JB. Prevalence of congenital optic disc pits in adult Chinese: The Beijing Eye Study. Eur J Ophthalmol. 2006; 16: 863-864.
4. Healey PR, Mitchell P. The prevalence of optic disc pits and their relationship to glaucoma. J Glaucoma. 2008; 17: 11-14.
5. Georgalas I, Ladas I, Georgopoulos G, Petrou P. Optic disc pit: a review. Graefes Arch Clin Exp Ophthalmol. 2011; 249: 1113-1122.
6. Stefko ST, Campochiaro P, Wang P, Li Y, Zhu D, Traboulsi EI. Dominant inheritance of optic pits. Am J Ophthalmol. 1997; 124: 112-113.
7. Lee PY, Huang MC, Wang TJ. Optic disc pit combining with an optic nerve cyst - A case report. Taiwan J Ophthalmol. 2012; 2: 31-4.
8. Bonnet M. Serous macular detachment associated with optic nerve pits. Graefes Arch Clin Exp Ophthalmol. 1991; 229: 526-532.
9. Wilkinson CP, Hinton DR, SriniVas RS, Wiedemann P. Ryan's Retina. Schachat AP (ed). 6th Edition. Elsevier, Amsterdam, The Netherlands, 2017.
10. Huang TL, Lin KH, Sheu MM, Tsai RK. Optical coherence tomography of a coexisting optic nerve cyst and optic disc pit: a case report. Kaohsiung J Med Sci. 2010; 26: 261-265.
11. Theodossiadis PG, Stringaris K, Papadopoulos V, Theodossiadis GP. Optic nerve cyst associated with optic disk pits. Graefes Arch Clin Exp Ophthalmol. 2005; 243: 718-720.
12. Ohno-Matsui K, Hirakata A, Inoue M, Akiba M, Ishibashi T. Evaluation of congenital optic disc pits and optic disc colobomas by swept-source optical coherence tomography. Invest Ophthalmol Vis Sci. 2013; 54: 7769-7778.
13. Nair AG, Kaliki S, Mishra DK, Naik MN. Unilateral optic disc pit associated with orbital cyst in a child. Saudi J Ophthalmol. 2017; 31: 190-192.
14. Vegunta S, Patel BC. Optic Nerve Cysts. StatPearls [Internet]. Available at: https://www.ncbi.nlm.nih.gov/books/NBK534104/, date accessed: 30/9/2024.