

## Optic disc pit maculopathy: one case report

Soukaina Tenouri<sup>1</sup>, Ahmed Bennis<sup>1</sup>, Fouad Chraïbi<sup>1</sup>, Meriem Abdellaoui<sup>1</sup>, Idriss Benatiya Andaloussi<sup>1</sup>

<sup>1</sup>(Ophthalmology Department, Hassan II University Center of Fez, Morocco)

---

### **Abstract:**

**Background:** Optic disc pit (ODP) is a rare congenital anomaly of the papilla. It is infrequent, seen in about 1 in 11 000 of the general population. Some patients develop an optic disc pit maculopathy (ODP-M) in which the visual acuity is lowered. ODP is congenital, but the development of ODP-M is unclear to date and can occur at any age.

**Case Report:** We report the case of a 35-year-old woman who consulted for blurred and distorted vision in the left eye. A fundus photograph of the left eye showed a typical optic disc pit. An optical coherence tomography (OCT) scan demonstrated multilayered retinoschisis. Thus, our diagnosis of optic disc pit maculopathy with retinoschisis was withheld. Given the good visual acuity (0.5 decimal scale), the therapeutic decision of follow-up was chosen.

**Conclusion:** ODP is a rare disease, and ODP-M treatment and management are difficult. The deficiency of suitable published data makes patient counseling and decision making even more intricate.

**Key Word:** Optic disc pit; Maculopathy; Congenital.

---

Date of Submission: 08-12-2021

Date of Acceptance: 23-12-2021

---

### **I. Introduction**

Optic disc pit (ODP) is a rare congenital anomaly of the papilla appearing as a grayish-white, round or oval depression at the optic nerve. It can also occur secondary to diseases such as glaucoma and myopia<sup>1,2</sup>. It is infrequent, seen in about 1 in 11 000 of the general population<sup>3</sup>, and there is no gender predilection<sup>4</sup>. Pits are generally unilateral but may be bilateral in up to 15% of patients<sup>5</sup>.

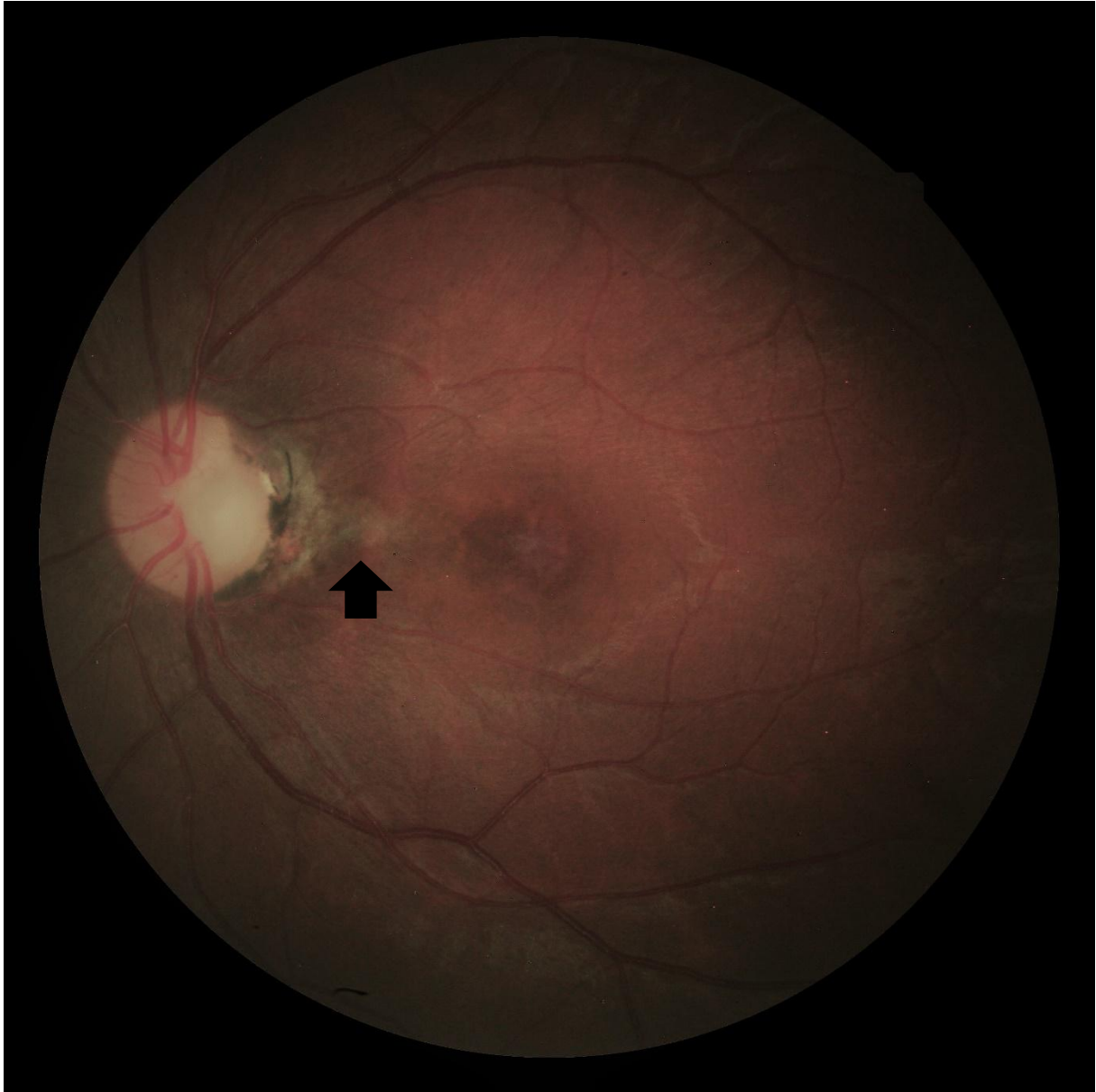
It has been estimated that approximately a quarter to three quarters of patients will develop serous detachment and/or retinoschisis of the central macula at some stage of their life, leading to the optic disc pit maculopathy (ODP-M)<sup>6,7</sup>. In such cases, the visual acuity is lowered. Otherwise, ODP is usually asymptomatic and may be observed incidentally.

ODP is congenital, but the development of ODP-M is unclear to date and can occur at any age.

### **II. Case Report**

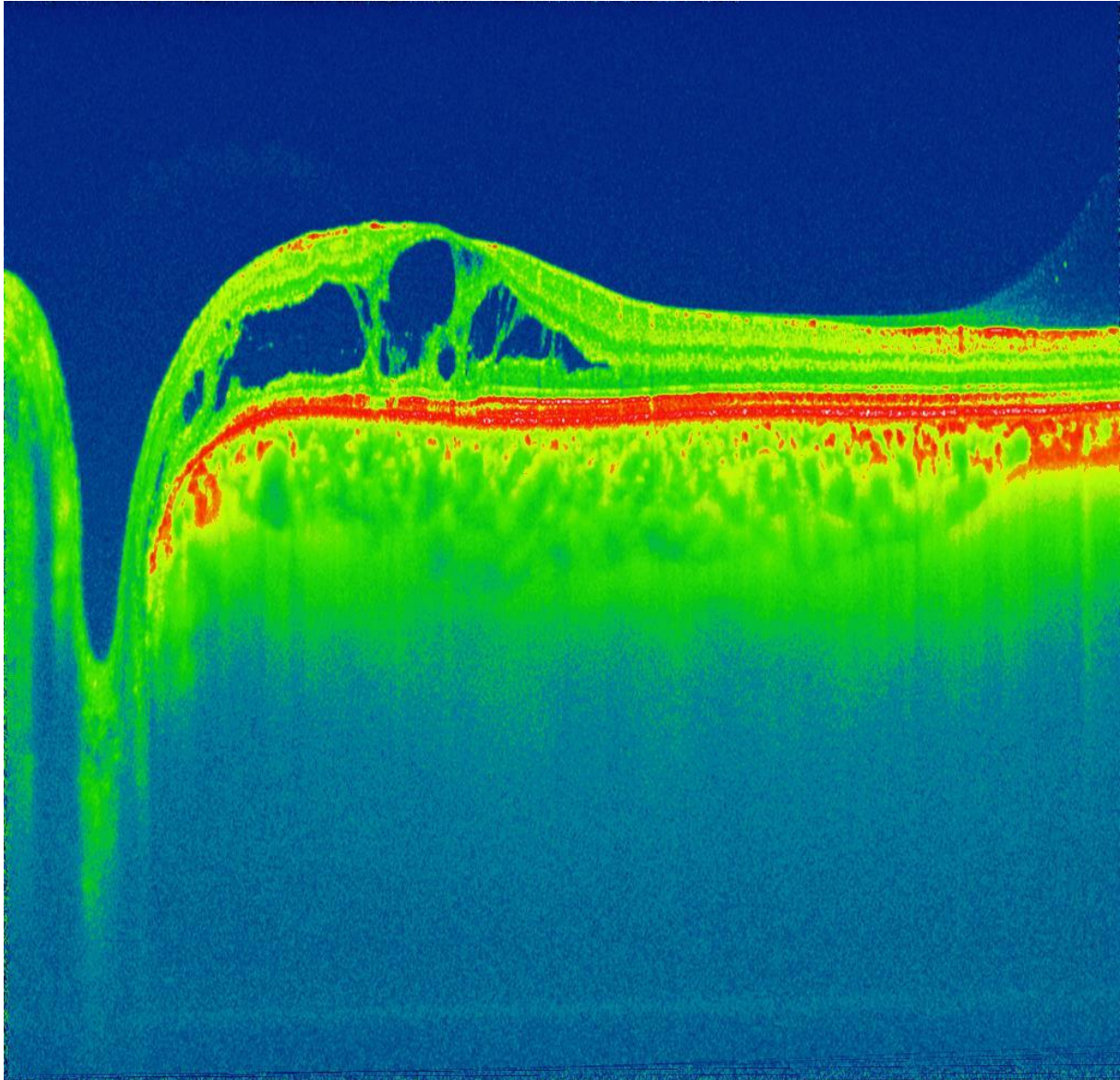
We report the case of a 35-year-old woman who consulted for blurred and distorted vision in the left eye for 03 months, with no associated pain or redness. She had no significant medical or ocular history.

Her visual acuity was: Right 1, Left 0.5 (decimal scale). Intra ocular pressure was normal at both eyes. Pupils were equal and reactive to light and the patient reported central distortion on viewing of the Amsler grid with the left eye. Figure 1 shows a fundus photograph of the left eye: the optic disc pit was visible as a round grey area at the temporal edge of the optic disc, without the presence of a retinal ciliary artery or arterial trifurcation, conventionally associated with this optic nerve anomaly.



**Figure 1:** Fundus photograph of optic disc pit (left eye). Arrow: optic disc pit.

An optical coherence tomography (OCT) scan (Figure 2) demonstrated multilayered retinoschisis. Thus, our diagnosis of optic disc pit maculopathy with retinoschisis was withheld. Given the good visual acuity, the therapeutic decision of follow-up was chosen, and the patient was educated on the importance of functional signs, regular fundoscopic examination and macular OCT controls.



**Figure 2:** Optical coherence tomography in color mode (left eye): multilayer retinoschisis in interpapillomacular area

### III. Discussion

An ODP appears as a localized grey (60%), white/yellow (30%), or black (10%) round or oval depression in the optic disc. This variation of color is attributed to the presence of glial tissue<sup>5,8</sup>. ODPs are most located at the inferotemporal segment of the optic disc, with 20% occurring centrally and 10% located in other regions of the optic disc. The size of the pit varies from 0.1 to 0.7 disc diameters<sup>9</sup>. ODPs may be associated with other abnormalities, such as nerve head coloboma and nerve head enlargement. In up to 60% of patients with ODP, one or two cilioretinal arteries can be seen emerging from the pit base<sup>5</sup>.

A review of the literature made on PUBMED with the keywords "optic disc pit" combined with "maculopathy" and "case" over a period of 20 years, ranging from 2000 to 2020, found 136 studies dealing roughly with the therapeutic aspect of pathology. A 2018 British study published in the American journal of ophthalmology identified 74 new cases of ODP-M over a 2-year study period<sup>10</sup>.

ODPs are usually asymptomatic and diagnosed as incidental findings during fundus examination. Visual deterioration occurs only when congenital ODPs are complicated by macular lesions such as macular edema, schisis-related macular detachment, or changes in macular pigment.

Although ODP is a congenital anomaly, no obvious triggering factors have been associated with the development of maculopathy<sup>11</sup>. ODP-M can occur at any age but typically is expected to occur in individuals during the third and fourth decades of life<sup>5,12</sup>. There are many theories regarding the pathophysiology of ODP-M. Some authors propose the intraretinal fluid is cerebrospinal fluid that leaks in via the ODP, other studies

suggest the fluid is liquefied vitreous that enters through a breach in the dysplastic retina overlying the ODP<sup>13,14</sup>.

The diagnosis of ODP-M is based mainly on fundus examination and OCT imaging. Fundus examination demonstrates an ODP that is usually located temporally with a coexistent macular elevation. OCT is the most helpful diagnostic tool since it reveals the detachment with a schisis cavity and with a coexisting outer layer detachment from the retinal pigment epithelium<sup>15,16</sup>. OCT may also reveal retinal edema in the inner layers, with cystic degeneration overlying a more central detachment<sup>9</sup>. A recent study showed that OCT can detect vitreous abnormalities in patients with ODP-M<sup>17</sup>.

At this time, various treatment modalities have been tried with variable success, and this is due to the rarity of the pathology and the challenging nature of the retinal status. The most widely accepted treatment is a surgical approach involving pars plana vitrectomy with or without internal limiting membrane peeling, with or without endolaser photocoagulation and C3F8 tamponade.

#### IV. Conclusion

ODP is a rare disease, and ODP-M treatment and management are difficult. The deficiency of suitable published data makes patient counseling and decision making even more intricate. Although spontaneous resolution can occur, the prognosis is generally poor and can lead to severe visual loss if left untreated.

Still the exact pathophysiology is still debated, management should be individualized, depending on the terrain. As knowledge of the disease increases, more methods of treatment will likely be developed. Therefore, better outcomes can be obtained.

#### References

- [1]. Javitt JC, Spaeth GL, Katz LJ, Poryzees E, Addiego R. Acquired pits of the optic nerve. Increased prevalence in patients with low-tension glaucoma. *Ophthalmology*. 1990 Aug;97(8):1038-43.
- [2]. Radius RL, Maumenee AE, Green WR. Pit-like changes of the optic nerve head in open-angle glaucoma. *Br J Ophthalmol*. 1978 Jun;62(6):389-93.
- [3]. Kranenburg Ew. Crater-like holes in the optic disc and central serous retinopathy. *Arch Ophthalmol*. 1960 Dec;64:912-24.
- [4]. Georgalas I, Ladas I, Georgopoulos G, Petrou P. Optic disc pit: a review. *Graefes Arch Clin Exp Ophthalmol*. 2011 Aug;249(8):1113-22.
- [5]. Brodsky MC. Congenital optic disk anomalies. *Surv Ophthalmol*. 1994 Sep-Oct;39(2):89-112.
- [6]. Brown GC, Shields JA, Goldberg RE. Congenital pits of the optic nerve head. II. Clinical studies in humans. *Ophthalmology*. 1980 Jan;87(1):51-65.
- [7]. Gordon R, Chatfield RK. Pits in the optic disc associated with macular degeneration. *Br J Ophthalmol*. 1969 Jul;53(7):481-9.
- [8]. Krivoy D, Gentile R, Liebmann JM, Stegman Z, Rosen R, Walsh JB, Ritch R. Imaging congenital optic disc pits and associated maculopathy using optical coherence tomography. *Arch Ophthalmol*. 1996 Feb;114(2):165-70.
- [9]. Reed D, David D. Congenital pits of the optic nerve. *Clinical Eye and Vision Care*. 1999 Nov;11(2):75-80.
- [10]. Steel DH, Suleman J, Murphy D C, Song A, Dodds S, Rees J. Optic disc pit maculopathy: a two-year nationwide prospective population-based study. *Ophthalmology*, 2018 *125*(11), 1757-1764.
- [11]. Sadun AA, Alfredo A, Khaderi KR, Khizer R. Optic disc anomalies, pits, and associated serous macular detachment. *Retina Fifth Edition*. Elsevier Inc. 2012. p. 1583-1588.
- [12]. Brockhurst RJ. Optic pits and posterior retinal detachment. *Trans Am Ophthalmol Soc*. 1975;73:264-91.
- [13]. Shah SD, Yee KK, Fortun JA, Albin T. Optic disc pit maculopathy: a review and update on imaging and treatment. *Int Ophthalmol Clin*. 2014 Spring;54(2):61-78.
- [14]. Moisseiev E, Moisseiev J, Loewenstein A. Optic disc pit maculopathy: when and how to treat? A review of the pathogenesis and treatment options. *Int J Retina Vitreous*. 2015 Aug 7;1:13.
- [15]. Chiu YT, Chen HY, Tsai YY, Lin JM, Chiang CC. Stratus optical coherence tomography for evaluating optic disc pits associated with maculopathy before and after vitrectomy: two case reports. *Kaohsiung J Med Sci*. 2006 May;22(5):229-34.
- [16]. Lincoff H, Schiff W, Krivoy D, Ritch R. Optic coherence tomography of optic disk pit maculopathy. *Am J Ophthalmol*. 1996 Aug;122(2):264-6.
- [17]. Theodossiadis PG, Grigoropoulos VG, Emfietzoglou J, Theodossiadis GP. Vitreous findings in optic disc pit maculopathy based on optical coherence tomography. *Graefes Arch Clin Exp Ophthalmol*. 2007 Sep;245(9):1311-8.

Soukaina Tenouri, et. al. "Optic disc pit maculopathy: one case report." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 20(12), 2021, pp. 25-28.