CASE REPORT

# OPTIC DISC PIT ASSOCIATED WITH CHOROIDAL COLOBOMA: CASE REPORT AND SHORT REVIEW

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Optic disc pits are hypothesized to form because of failure of embryonic fissure closure, which can also present with congenital defects in the choroid, RPE, and neurosensory retina. It is also associated with serous macular detachment. We present a case report of a 32-year-old man with an optic disc pit and independent choroidal coloboma below the inferior peripapillary area in the left eye, associated with macular retinoschisis with serous detachment.

**Keywords:** Coloboma; Optic disc pit; Choroid; Serous detachment

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## INTRODUCTION

The optic disc pit is an uncommon abnormality of the optic nerve head, which has been hypothesized to probably arise because of defective closure of the superior end of the embryonic fissure.[1] It may be either acquired or congenital and can cause loss of vision and serous macular detachment.[1] Most commonly the pit appears unilaterally and in the inferotemporal region of the optic disc, and it is greyish oval/round in appearance. In the initial phase, the optic pit is hypo-fluorescent on fundus fluorescein angiography (FFA); though, the late phase shows hyper-fluorescence.[2]

Incidence of congenital optic disk pits is rare, as it can occur in about one in 11,000 people and occur equally in men and women and thus are extremely rare.[3] Serous macular detachment can occur in association with optic nerve pits or similar congenital cavitary disc anomalies, or with chorioretinal coloboma.[3] In this article, we present a rare case of an optic disc pit with central serous chorioretinopathy and coexistent independent choroidal coloboma.

## CASE PRESENTATION

A 32-year-old male patient came to the outpatient department (OPD) clinic in August 2019 at LRBT Tertiary and Teaching Eye Hospital (Karachi) with the complaint of sudden painless decreased vision in his left eye. Informed consent was obtained from the patient. His bilateral adnexal examination was unremarkable, and no significant changes in anterior segment examination were identified. Fundus examination of the right eye showed no changes in the macula with normal foveal reflex and optic disc: however, the left eye revealed an oval and grey inferotemporal invaginated defect on the optic disc. There was a yellowish flat circumscribed lesion about one-fourth diameter of the optic disc, located half-disc size in the inferior peripapillary area. The foveal reflex was dull with circumscribed elevation at the macula. Spectral-domain Optical coherence tomography (SD-OCT) of the macula displayed serous detachment with optic disc pit and choroidal coloboma as shown in figure-2.

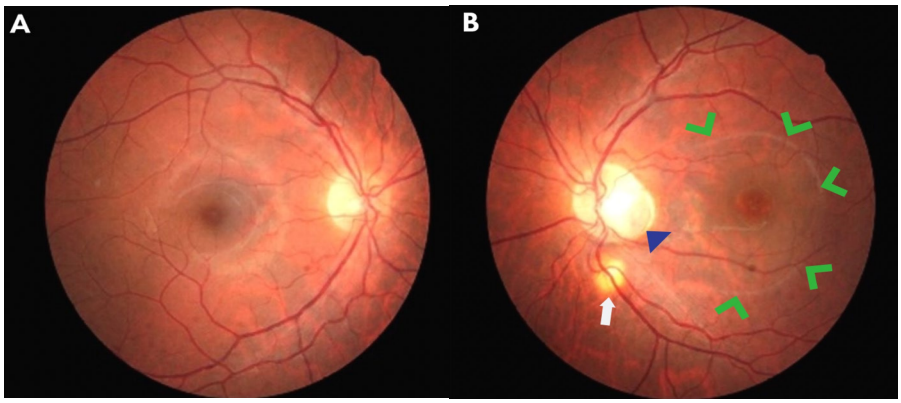


**Figure-1: Fundus photograph of the right eye (A) showing normal optic disc with good macular reflex. Fundus photograph of the left eye (B) shows an optic disc with an excavated defected with an invagination on the inferotemporal surface of the disc corresponding to an optic disc pit (blue arrowhead). A choroidal coloboma can be seen inferior to the disc in the inferior peripapillary region (white arrow). The foveal reflex is dull with circumscribed elevation at the macula depicting serous detachment (green arrowheads).**

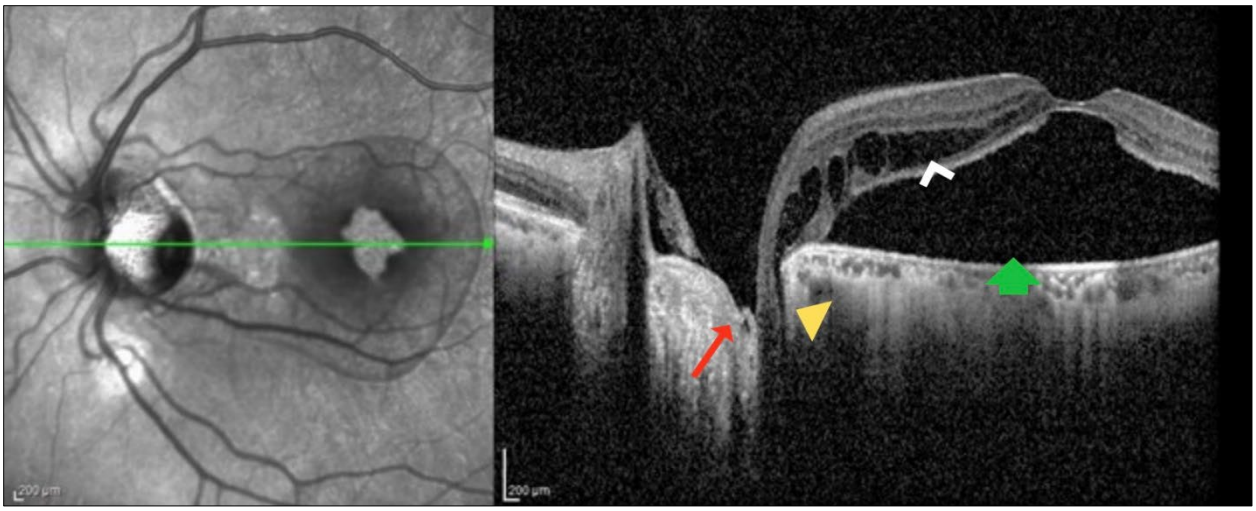

**Figure-2: Spectral domain OCT (SD-OCT) of the left eye. An optic pit (red arrow) is seen along with a communication (yellow arrowhead), causing serous detachment. Subretinal fluid (green arrow) and oedema (white arrow) can be visualized between the layers of the retina**.

**Table-1: Summary of optic disc pits associated with the optic disc, iris, retinal, lens or choroidal colobomas reported to date**

| Author (Year) | Age | Gender | Eye involved | Optic disc pit location | Coloboma type and location | Other associations |
|---|---|---|---|---|---|---|
| Brown and Jerry *et al*. (1980) | N/A | N/A | N/A | Temporal | Inferior optic disc | |
| | N/A | N/A | N/A | N/A | Optic disc and choroid | Retinal pigment epithelium |
| | N/A | N/A | N/A | N/A | Optic disc | |
| Brown and Ausberger (1980) | 45 | M | Both | Inferonasal | Retino-choroidal | |
| Singerman and Mittra (2001) | N/A | N/A | N/A | Inferior | Iris | |
| | N/A | N/A | N/A | Inferior | Iris | |
| | N/A | N/A | N/A | Inferior | Iris | |
| Saatci *et al* (2004) | 48 | F | Left | Inferotemporal | choroidal | |
| D. Cruzado-Sánchez *et al.* (2013) | 38 | F | Left | Temporal | Choroidal | Serous macular detachment |
| Ozelce *et al.* (2016) | 42 | F | Right | Infero-temporal | Iris, lens and 2 choroidal | |
| | | | Left | - | Iris and lens | |
| | 21 | M | Left | Infero-temporal | Choroidal | |
| Takkar B *et al.* (2017) | 6 | M | Right | Inferior | Optic disc | Retinoschisis |
| | | | Left | - | Choroidal | |
| Storch *et al*. (2017) | 33 | F | Left | Infero-temporal | Iris-Retino-Choroidal | |
| Ricardo *et al*. (2019)[9] | 21 | F | Left | Infero-temporal | Optic disc | Macular retinoschisis |
| Sachan *et al.* (2021) | 18 | M | Right | Inferior | Choroidal | Multiquadrant peripapillary retinoschisis |

## DISCUSSION

The pathophysiology behind the development of optic disc pit is ambiguous. Two hypotheses have been proposed: either aberrant development of the primal papilla of optic-nerve and incomplete resolution of peripapillary neuroectodermal folds[4] or the incomplete closure of embryonic fissure[5–7].

A handful of cases have been reported of pits in the optic nerve head co-existing with choroidal, retinal-choroidal coloboma, both unilaterally[6,8–11] and bilaterally[2,7,12,13], summarized in table-1. Only three cases with significant inferior optic disc colobomas, one of which also had an inferior colobomatous defect of the choroid and retinal pigment epithelium (RPE), were recorded in Brown's analysis of 75 eyes with congenital optic nerve pits.[2] Singerman and Mittra described a peculiar case in a family of three generations showing classic optic pits and iris colobomas.[5] Embryonic fissures that fail to close, can lead to defects in the choroid, RPE, and neurosensory retina extending along a line from the nerve to the iris inferonasal.[6] Combinations of these cavitary defects can appear in a single patient such as in the case published by Özelce *et al*. showing optic disc pit co-

occurring with lens, iris and choroidal colobomas and another case with co-existent choroidal coloboma, supporting the theory that inadequate closure of the embryonic fissure being the cause of optic pits.[5,6,13] Overall, only a small number of cases illustrating the coexistence of an optic disc pit and a choroidal coloboma have been documented to our knowledge.[2,5,6,8,10–13]

25–75% of eyes in optic pits develop serous macular detachments, usually causing central vision loss in the third or fourth decade of life.[3] It is possible for ophthalmologists to monitor individuals with these embryologic anomalies closely and diagnose serous maculopathy earlier if these defects are identified early when the patients are asymptomatic. A submacular fluid collection can cause a serous detachment, peripapillary chorioretinal atrophy, changes in the retinal pigment epithelium, and neovascularization of peripapillary subretinal space, if not treated early.[6]

Our case report describes an unusual finding of choroidal coloboma with an optic disc pit in the left eye with an associated serous macular detachment, in a patient with a complaint of sudden painless loss of vision. To conclude, this case study emphasizes the importance of early diagnosis and monitoring of individuals with congenital cavitary abnormalities such as optic pit and coloboma to improve the prognosis for their vision. To our knowledge, this is the first case documented of an optic pit co-existent with a choroidal coloboma in our population.

## AUTHOR CONTRIBUTIONS

MUJ and SFR conceived the idea and designed the study outline. MUJ and AM wrote the draft of the manuscript, and prepared figures, and a table of the manuscript. AM performed the literature review and improved the manuscript. SFR supervised the project and did the final proofreading. All authors contributed to the article and approved the submitted version.

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**Conflict of Interest:** None to declare.
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