Coexistence of an Optic Pit within a Chorioretinal Coloboma leading to Retinal Detachment

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Abstract

INTRODUCTION:
Congenital cavitary anomalies of the optic disc and retina are an uncommon cause of vision loss and often go unrecognized. The most common presenting complaint in patients with an optic pit or coloboma is gradual and progressive monocular vision loss. Asymptomatic identification and close monitoring of these congenital anomalies is important because 25-75% of eyes with an optic pit will develop a serous macular detachment leading to vision loss by the third or fourth decade of life. This is a case of a patient with bilateral chorioretinal coloboma with a coexistent optic pit within the coloboma in the left eye leading to a localized retinal detachment.
METHODS:
An adolescent male was referred for vitreoretinal evaluation of monocular vision loss. His visual acuity was 20/50 OD and hand motion OS with normal IOP. He has a past medical history of intellectual disability with no other neurologic abnormalities. Anterior segment, ocular motility, and pupillary exams were normal. Dilated fundus examination of the right eye revealed an inferior typical retinochoroidal coloboma without involvement of the disc. The left eye revealed an optic pit within an inferior retinochoroidal coloboma along with a localized retinal detachment temporal to the coloboma.
RESULTS:
Our case is a rare example of an optic pit located inside a coexistent chorioretinal coloboma, that has not been previously described in the literature and provides support for the embryonic fissure theory of coloboma and optic pit development.
CONCLUSION:
This case report underlines the importance of early identification and monitoring of patients who demonstrate congenital cavitary anomalies of the retina like optic pit and colobomas due to the potential for a devasting visual outcome if not observed and prompted treated. To our knowledge, this is the first case documented of an optic pit located inside of a chorioretinal coloboma.

Introduction

Congenital cavitary anomalies of the optic disc and retina are an uncommon cause of vision loss and often go unrecognized [1,2]. The most common presenting complaint in patients with an optic pit or coloboma is gradual and progressive monocular vision loss [2]. Patients are often referred for neurological evaluation of their vision loss because the congenital defect has not been identified previously [2]. The cause of visual impairment from optic pits and colobomas is from central serous retinopathy, macular edema, or retinal detachment. The source of the subretinal fluid leading to the vision loss has not been clearly established [2,3]. Patients can also display visual field defects manifesting as an arcuate defect or enlarged blind spot.

Optic pits are oval depressions of the optic disc that are composed of pigment epithelium with neuroretinal and glial elements [2,4]. Histologically, the retinal elements herniate into a collagen lined pocket extending posteriorly into the subarachnoid space through a defect in the lamina cribrosa [3,5]. Optic pit is rare with an incidence of 1 in 11,000 and a prevalence between 0.02-0.19% [4,6]. It is unilateral in 85% of cases, most occuring in the inferotemporal border of the optic disc [2,3,4]. Optic pits are not hereditary and are not known to be associated with other systemic or central nervous system malformations, while colobomas are [2,3,6].

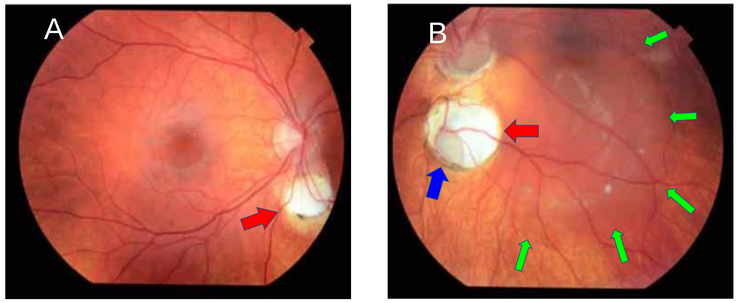
Colobomas are another congenital cavitary anomaly of the optic disc, choroid, retina, or iris that result from partial or complete failure of the embryonic optic fissure [6,7]. Coloboma are often isolated, but can be associated with systemic abnormalities like CHARGE syndrome or other midline abnormalities [2,7]. Multiple generations of the same family have been reported to have coloboma showing that coloboma may be inherited in an autosomal dominant pattern [2,8]. Coloboma are slightly more common than optic pits with an incidence between 0.5-2.2 cases per 10,000 live births [7]. Unlike optic pits, coloboma occurs bilaterally just as frequently unilaterally [2].

Coexistence of optic pit and choroidal coloboma is rarely encountered [1]. There have been seven cases of coexisting optic pit and coloboma reported in the literature [1,6,8-12]. Asymptomatic identification and close monitoring of these congenital anomalies is important because 25-75% of eyes with an optic pit will develop a serous macular detachment leading to vision loss by the third or fourth decade of life [3,4]. The authors report a case of a patient with bilateral chorioretinal coloboma with a coexistent optic pit within the coloboma in the left eye leading to a localized retinal detachment.

Patient History

An adolescent male was referred to the vitreoretinal service for ophthalmologic evaluation of monocular vision loss. His visual acuity was 20/50 OD and hand motion OS with intraocular pressures of 16 OU. Refraction plano. Visual fields were full to confrontation in both eyes. He has a past medical history of intellectual disability, with no other systemic or neurologic abnormalities. Anterior segment biomicroscopy, extrinsic ocular motility, and pupillary light reflex were normal. Dilated fundus examination of the right eye revealed an inferior typical retinochoroidal coloboma without involvement of the disc (Figure 1a). The left eye revealed an optic pit within an inferior retinochoroidal coloboma along with a localized retinal detachment temporal to the coloboma (Figure 1b). Our case is a rare example of an optic pit located inside a coexistent chorioretinal coloboma. This observation has not been previously described in the literature and provides more support for the theory that optic pits may be due to the defective closure of the embryonic optic fissure.

Fundus Photography



A. Fundus photo of the right eye revealing a normal appearing optic disc with a chorioretinal coloboma (red arrow) inferior to the optic nerve head and independent of the disc.

B. Fundus photo of the left eye reveals a hypoplastic optic disc with a chorioretinal coloboma (red arrow) inferior to the optic nerve head and independent of the disc. Within the coloboma there is an oval-shaped gray depression inferiorly which is consistent with an optic pit (blue arrow). There is also a localized serous detachment (green arrows mark borders) involving the macula, temporal aspect of the coloboma, and inferotemporal aspect of the disc.

Discussion

The embryologic origin of optic pits is debated in the literature. One theory is that optic pits are thought to develop by anomalous development of the primordial optic nerve; however, others favor a theory, similar to the origin of colobomas, focusing on an incomplete closure of the embryonic fissure [1]. During development, the optic cup is created as the distal portion of the primary optic vesicle approaches the proximal portion after invagination. Abnormal closure in different locations during optic cup formation will determine the location of the cavitary abnormality. A chorioretinal coloboma is formed when there is abnormal closure in the inner layer of the optic up; whereas, incomplete closure of the distal end of the embryonic fissure causes an iris coloboma [11].

Optic pits are thought to develop by anomalous development of the primordial optic nerve [1,5,6,8]; however, others favor a theory focusing on the incomplete closure of the embryonic fissure [1,5,8,11]. The anomalous growth theory suggests an impaired differentiation of primitive peripapillary sclera from the primary mesenchyme causes the formation of optic pits [5,6]. This theory is supported by a rare association with retinochoroidal colobomas [8]. The abnormal differentiation would also lead to a communication between the pit and the underlying subarachnoid space [6]. The embryonic fissure theory, discussed above in coloboma formation, is supported by the presence of defects inferonasally extending from the nerve to the iris due to failure of embryonic fissure to close [1,8,11]. This may also cause defects in the choroid, RPE, and neurosensory retina along a line extending inferonasally from the optic disc to the pupillary margin of the iris [1]. Combinations of these cavitary defects can occur in a single patient [2],such as the coexistence of optic pits and uveal colobomas reported by Ozelce, further supports the hypothesis of incomplete closure of the embryonic fissure being the etiology of optic pits[11].

Optic pits and optic colobomas are important to recognize on ophthalmic examination as their sequelae can be devastating to vision. Serous macular detachments develop in 25-75% of eyes with optic pits. These detachments often occur in the 3rd or 4th decade of life which leads to central vision loss [3,4]. This underscores the importance of early detection of these embryonic cavitary defects while patients are asymptomatic, as well as subsequent continued monitoring after initial detection in order to address and promptly treat serous maculopathy. Delayed treatment of the submacular fluid can lead to a serous detachment, peripapillary chorioretinal atrophy, RPE changes, and peripapillary subretinal neovascularization [1].

Conclusions

In conclusion, this case report underlines the importance of early identification and monitoring of patients who demonstrate congenital cavitary anomalies like optic pit and coloboma. The visual prognosis is positive if these abnormalities are observed and patients are educated about the potential complications. These embryological abnormalities are rare and to our knowledge, this is the first case documented of an optic pit located inside of a chorioretinal coloboma.

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