Case Report

Bilateral optic nerve head drusen with chorioretinal coloboma in the right eye

Ali Reza Dehghani, Leila Rezaei[1], Heshmatollah Ghanbari

Isfahan Eye Research Center, Isfahan University of Medical Sciences, Isfahan, [1]Emam Khomeini Eye Research Center, Kermanshah University of Medical Sciences, Kermanshah, Iran

Abstract

Chorioretinal coloboma is a congenital defect of the eye caused by improper closure of the embryonic fissure. Optic nerve head drusen (ONHD) are white calcareous deposits that are generally asymptomatic. We report a very rare association of both in a healthy patient with no any systemic syndrome. A 16-year-old man was referred to our clinic from suffering blurred vision. Best corrected visual acuity of the right eye was 6/10 and 10/10 in the left one. External ocular and slit lamp examination were normal. Dilated ophthalmoscopy showed marked swelling in both optic nerves and chorioretinal coloboma in the right eye inferiorly. Ultrasonography showed an echodense structure with acoustic shadowing in both eyes consistent with buried ONHD. Visual field testing showed normal field in the left eye and moderate superior field depression in the right eye corresponding to inferior coloboma in funduscopy. Results of general medical and neurologic, cardiologic, and other examinations were normal. To the best our knowledge combination of bilateral ONHD and unilateral chorioretinal coloboma in a healthy patient with no any systemic syndrome has not been published in the literature. We reported this very rare association and recommended examine eyes and other body organs. In such cases that coloboma is associated with ONHD, we should keep in mind Noonan syndrome. The diagnosis of Noonan syndrome is clinical and confirm by the consultant pediatricians and clinical geneticists.

Key Words: Coloboma, optic nerve head drusen, retinal detachment

Address for correspondence:
Dr. Leila Rezaei, Emam Khomeini Eye Research Center, Kermanshah University of Medical Sciences, Kermanshah, Iran. E-mail: leyla_rezaei60@yahoo.com
Received: 03.03.2015, **Accepted:** 19.08.2015

INTRODUCTION

Coloboma results from defective closure of the embryonic fissure of the optic cup. These defects are typically located in the lower part of the iris, chorioretina, or optic disc. The typical coloboma occurs in the inferonasal quadrant, caused by the defective closure of the fetal fissure. Coloboma is identified by careful ophthalmoscopic examination and photography of both fundus of the presenting individual and their family members.[1] There is no treatment, and they may be complicated by glaucoma, retinal detachment, and central serous retinopathy.[2]

Optic nerve head drusen (ONHD) are incidental ophthalmologic finding in the optic nerve. Patients with ONHD are often asymptomatic, but sometimes present with a transient visual

Access this article online	
Quick Response Code:	Website: www.advbiores.net
	DOI: 10.4103/2277-9175.178798

How to cite this article: Dehghani AR, Rezaei L, Ghanbari H. Bilateral optic nerve head drusen with chorioretinal coloboma in the right eye. Adv Biomed Res 2016;5:34.

loss in 8.6% of patients. ONHD are of two types: Superficial and deep. The deep-buried drusen mimic papilledema. Because of the varied presentation deep-buried drusen pose a diagnostic challenge to the ophthalmologists. In young patients, they are mistaken for papilledema as it is clinically difficult to detect a buried drusen in the optic nerve head. They are observed as pale yellow lesions in the optic nerve head. Clinical examination aided by diagnostic tests like ultrasound B-scan can help establish the diagnosis.[3]

We report a patient with a combination of ONHD and coloboma. To the best our knowledge, association of chorioretinal coloboma and ONHD in the healthy patient has not been published in the literature.

CASE REPORT

A 16-year-old man was referred to our clinic from suffering blurred vision of right eye since childhood.

The patient had no past medical and familial history of any ophthalmologic disease or refractive errors.

There was no history of trauma, eye pain, glaucoma, double vision, headache, vertigo, dysarthria, tremor, paralysis, or loss of sensation. The patient did not have diabetes mellitus, hypertension, or other cardiovascular disease.

Ocular examination revealed an uncorrected visual acuity of 4/10 in the right and 10/10 in the left eye. Best corrected visual acuity of the right eye was 6/10 due to a refractive amblyopia. His cycloplegic refraction were –0.50 diopter sphere and –2.50 × 180 cylinder in the right eye and +0.75 diopter sphere and –0.50 ×59 cylinder in the left one.

The pupil's reaction to light, external ocular motility, palpebral fissures, the angle of the palpebral fissures, cranial nerves, stereoacuity, and exophthalmometry were normal, with no ptosis and strabismus. There was no afferent pupillary defect. Slit lamp examination was normal. The corneal sensation was intact.

Intraocular pressures measured using Goldman applanation tonometery were 11 mmHg in the right eye and 13 mmHg in the left eye.

Dilated ophthalmoscopic examination showed marked swelling in both optic nerves and diffuse elevation of the optic nerve and a large chorioretinal coloboma in the right eye inferiorly [Figures 1 and 2].

Ultrasonography with 10-MHz probe revealed echodense structure with acoustic shadowing in both eyes consistent with buried optic disc drusen [Figure 3].

24–2 visual field testing showed normal field in the left eye and moderate superior field depression in the right eye corresponding to inferior coloboma in funduscopy [Figures 4 and 5].

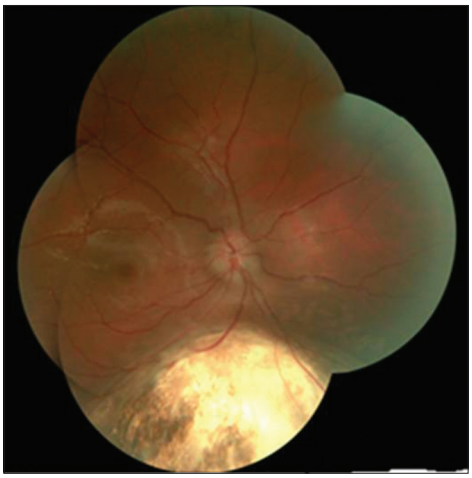

Figure 1: Wide field fundus photograph shows marked swelling of optic nerves and a large chorioretinal coloboma in the right eye inferiorly

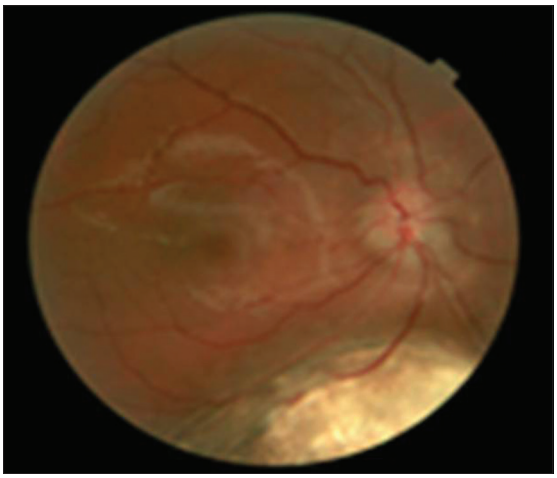

Figure 2: Fundus photograph shows optic nerve swelling and a large chorioretinal coloboma in the right eye

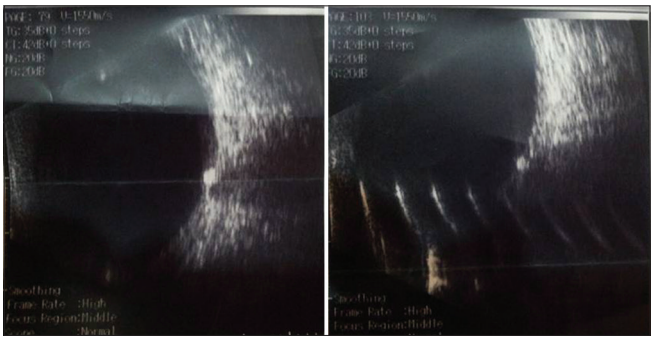

Figure 3: Ultrasonography shows echodense structure with acoustic shadowing in both eyes consistent with buried optic disc drusen

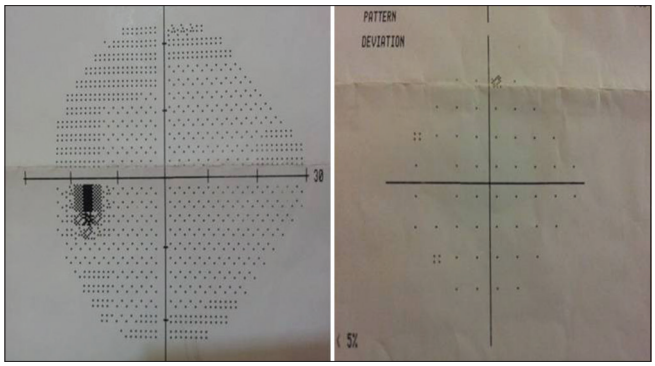

Figure 4: Visual field of left eye is normal

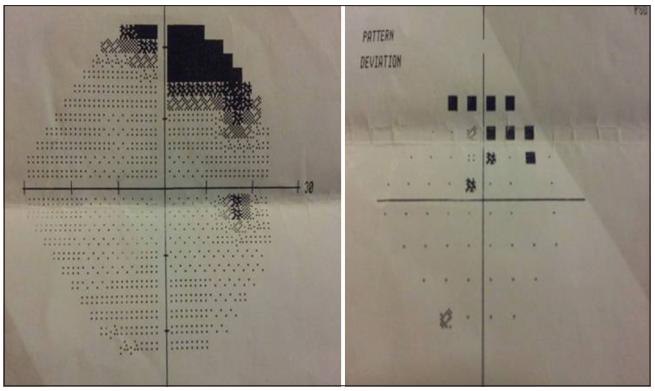

Figure 5: Visual field testing shows moderate superior field depression in the right eye corresponding to inferior coloboma in funduscopy

Consultant pediatrician, a pediatric cardiologist, neurologist, and clinical geneticist visited our patient and his family members. Results of general medical and neurologic, cardiologic and other examinations performed were normal.

Results of standard laboratory testing, pneumoencephalography, electroencephalography, and brain magnetic resonance imaging were unremarkable.

DISCUSSION

ONHD are white calcareous deposits that are generally asymptomatic and are bilateral in 66–85% of cases.[4] They are characteristically found in the prelaminar region of the ONH and are believed to arise from the debris due to long standing axonal stasis in the nerve fiber layer.[5] Histopathologically, the drusen contain multiple deposits of calcium crystals that vary in size (5–1000 microns).[6,7]

A high index of suspicion is needed when young patients present with disc edema. Ultrasound B-scan is a sensitive tool to detect ONHD.[8]

Coloboma is a congenital defect of the eye caused by improper closure of the embryonic fissure. This condition occurs in 0.14% of the general ophthalmic population.[9]

Normal ocular development entails a series of tightly choreographed events, of which fusing the edges of the optic cup's embryonic fissure represents a key step in forming the future spherical eye. Disruption of this process results in cavitary anomalies affecting ocular tissues.[10]

Congenital cavitary anomalies of the optic disc, including coloboma, optic pit, morning glory anomaly, and extrapapillary cavitation.[10]

A variety of systemic and neurologic conditions may accompany these optic disc anomalies. These anomalies are associated with maculopathy and serous detachment. The unifying anatomic theme of these anomalies is the presence of a scleral (or lamina cribrosa) defect permitting anomalous communications between intraocular and extraocular spaces. These communications enable the critical pathogenic mechanism responsible for the maculopathy, namely, and dynamic fluctuations in the gradient between intraocular and intracranial pressures that direct the movement of fluid (vitreous humor or cerebrospinal fluid) into and under the retina.[11]

Also, morning glory syndrome can be associated to optic nerve coloboma or optic nerve drusen. This anomaly is characterized by an increase in optical disc size with poor defined wedges and involved by pigmented ring, deep excavation, the presence of glial tissue filling the optical disc, and radial disposition of retinal vessels.[12]

Interestingly, we were not able to find any previous report of combined bilateral ONHD and unilateral chorioretinal coloboma in a healthy patient with no any systemic syndrome in the literature.

This patient need close follow-up and three important points are necessary:

- Our patient also had chorioretinal coloboma in the right eye. Retinal detachment is the most common complication associated with chorioretinal coloboma, due to breaks within or adjacent to the coloboma. The retina overlying the choroidal defect in such cases remains thin and undifferentiated and, therefore, prone to retinal breaks and detachment. Most retinal detachments associated with chorioretinal coloboma require surgery and have poor visual outcomes. Forty percent of affected individuals may develop retinal detachment during their lifetimes.[9-15] It is recommended to examine eyes affected by chorioretinal coloboma at least annually to prevent

complications such as retinal detachment.[10] If a barrier was incomplete or missing, one could be enforced with a laser to limit either detachment or the risk thereof to one side of the margin

- ONHD can also cause anterior ischemic optic neuropathy (AION) due to crowding and compression on the axons.[16] It can be a rare cause of AION in young patients.[8] A high index of suspicion is needed when young patients present with disc edema and loss of vision, in such patient could have avoided the unnecessary neurological workup[3]
- In such cases that coloboma is associated with ONHD, we should keep in mind Noonan syndrome. Noonan syndrome is a genetic condition inherited in an autosomal dominant manner, characterized by heart disease, short stature, abnormal faces, and the somatic features of Turner's syndrome, but a normal karyotype. External features were hypertelorism, downward sloping palpebral apertures, epicanthic folds, ptosis, strabismus, and nystagmus. Anterior segment changes consisting of: Prominent corneal nerves, anterior stromal dystrophy, cataracts, and panuveitis.[17,18] Fundus changes including ONHD, optic disc hypoplasia, coloboma, and myelinated nerves. The diagnosis of Noonan syndrome is made purely on clinical grounds. The diagnosis of Noonan syndrome was confirmed by the consultant pediatricians and clinical geneticists.[18,19]

Results of general medical and neurologic, cardiologic and other examinations in this patient were normal, and Noonan syndrome be ruled out.

CONCLUSION

The combination of bilateral ONHD and unilateral chorioretinal coloboma in a healthy patient with no any systemic syndrome has not been published in the literature. We reported this rare association and recommended to examine eyes and other body organs. In such cases that coloboma is associated with ONHD, we should keep in mind Noonan syndrome. The diagnosis of Noonan syndrome is clinical and confirm by the consultant pediatricians and clinical geneticists.

Acknowledgments

The authors wish to thank all the colleagues who supported the research, especially Shahla Rezaei and Maryam Ghalyani for assistance with editing of the article.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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