A Case with Bilateral Chorioretinal Coloboma Appearing as Double Optic Disc

Çift Optik Disk İzlenimi Veren Bilateral Koryoretinal Kolobom Olgusu

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ABSTRACT Choroidal coloboma is a congenital anomaly caused by incomplete and defective closure of the embryonal fissure. Choroidal colobomas are frequently associated with some ocular and systemic anomalies involving the cardiovascular, central nervous, musculoskeletal, gastrointestinal, genitourinary and nasopharyngeal systems. In this article, we report a case of bilateral chorioretinal coloboma mimicking accessory optic disc. Thirty four year-old woman visited us with a complaint of low visual acuity in both eyes and strabismus. At ophthalmological examination, her best-corrected visual acuity was 2/10 with (+10.75–5.00x90˚) on right eye and 2/10 with (+11.00-3.75x 55˚) on left eye. 35-40 PD of exotropia was detected with prism alternating cover test. Fundus examination revealed bilateral chorioretinal coloboma. Fluorescein angiography revealed no notable findings other than chorioretinal coloboma. Extraocular muscles, optic nerve (single optic nerve in each orbit) and retrobulber structures were normal in both eyes under magnetic resonance imaging evaluation.

Key Words: Coloboma; optic disk; choroid

ÖZET Koroidal kolobom embriyonal yarığın tam olmayan veya hatalı kapanması sonucu oluşan konjenital bir anomalidir. Koroidal kolobomlar sıklıkla kardiovasküler sistem, merkezi sinir sistemi, kas-iskelet sistemi, gastrointestinal sistem, genitoüriner sistem ve nazofaringeal sistemleri de içerebilen sistemik ve oküler anomaliler ile birliktelik gösterirler. Bu yazıda, her iki tarafta aksesuar optik disk görünümünde olan koryoretinal kolobomlu bir olgumuzu paylaştık. Otuz dört yaşında kadın hasta her iki gözde az görme ve şaşılık şikayeti ile kliniğimize başvurdu. Oftalmolojik muayenede, en iyi düzeltilmiş görme keskinliği sağ gözde 2/10 (+10,75–5,00x90˚tashih ile), sol gözde 2/10 (+11,00-3,75x55˚tashih ile) idi. Alternan prizma örtme testi ile 35-40 PD bir ekzotropya tespit edildi. Fundus muayenesinde, bilateral koryoretinal kolobom görüldü. Floresein anjiyografide, koroidal kolobom dışında anlamlı bir değişiklik görülmedi. Manyetik rezonans görüntülemesinde, ekstraoküler kaslar, optik sinir (her orbitada bir adet optik sinir) ve retrobulber yapılar her iki gözde doğal görünümde idi.

Anahtar Kelimeler: Kolobom; optik disk; koroid

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Choroidal coloboma is a congenital anomaly caused by incomplete and defective closure of the embryonal fissure.[1] The colobomatous area lacks choroid and retina pigment epithelium. In some cases, although the retina is present, it is hypoplastic and gliotic.[2] Defect size and location varies. It is a rare malformation occuring in 0.14 % of the general population.[3] It can be unilateral or bilateral. 60% of the cases are bilateral.[4] Choroidal colobomas are frequently associated with some ocular and systemic anomalies involving the cardiovascular, central nervous, muscu-

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loskeletal, gastrointestinal, genitourinary and nasopharyngeal systems.[5] In this article, we report a case with bilateral chorioretinal coloboma mimicking accessory optic disc.

CASE REPORT

A 34 year-old woman visited us with a complaint of low visual acuity in both eyes and strabismus. On ophthalmological examination, her best-corrected visual acuitywas 2/10 with (+10.75–5.00 x 90˚) on right eye and 2/10 with (+11.00-3.75x 55˚) on left eye. 35-40 PD of exotropia was detected with prism alternating cover test performed with glasses and alternance was present. There was not notable findings regarding the anterior segment under slit lamp examination. The light reflex was normal in both eyes, and relative afferent pupillary defect was not seen. There was not any restriction of eye movements. Fundus examination revealed bilateral chorioretinal coloboma. Retina was attached and optic discs were non-colobomatous with less than 0.1 cup to disc ratio in both eyes. On right eye, choroidal coloboma was located on one and a half optic disc diameter inferior to the optic disc. In left eye, it was located on one disc diameter inferior to the optic disc. The size of the choroidal coloboma was half of the optic disc in right eye and same size with the optic disc in left eye (Figures 1, 2). Fundus fluorescein angiography revealed no notable findings other

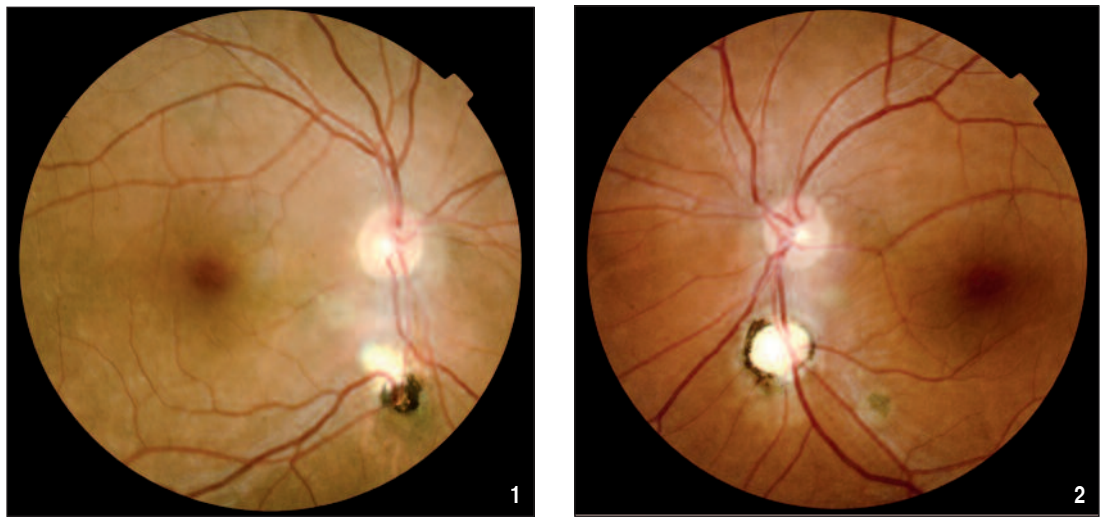


FIGURE 1, 2: Color photographs of fundus showing chorioretinal coloboma in both eyes.

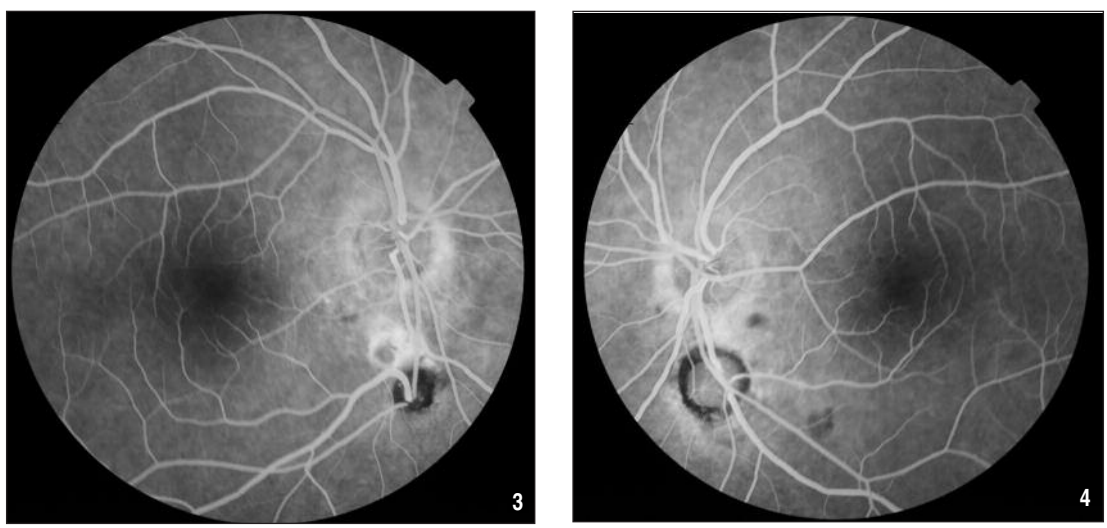
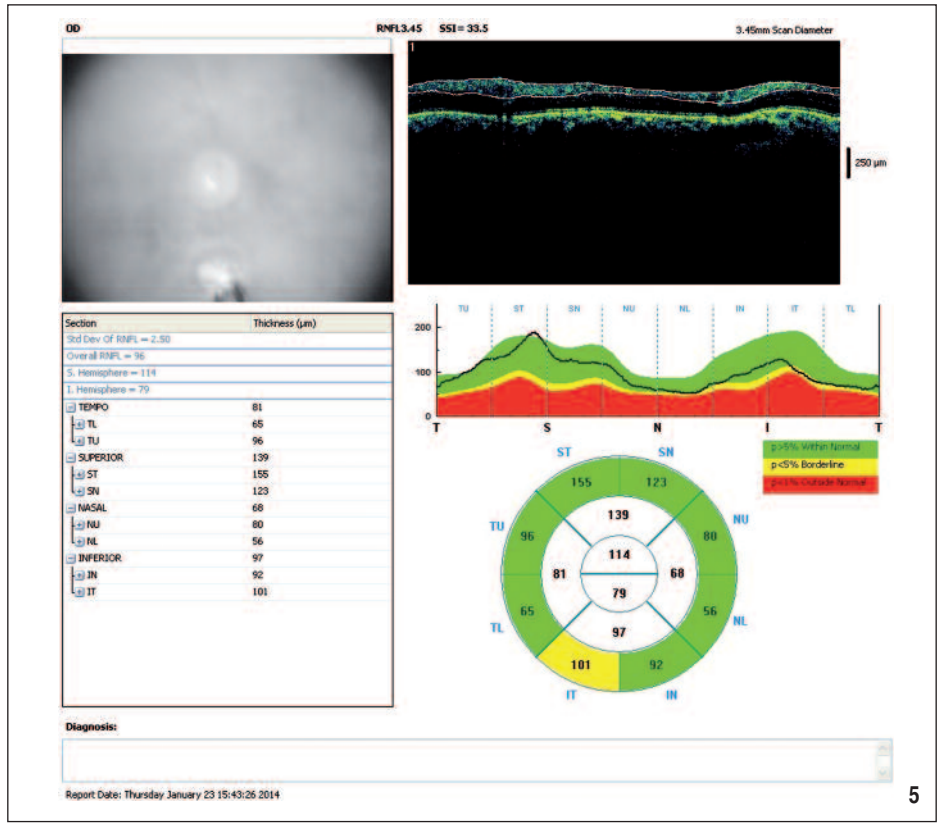


FIGURE 3, 4: Fundus fluorescein angiography of the patient showing only coloboma without other retinal lesions.



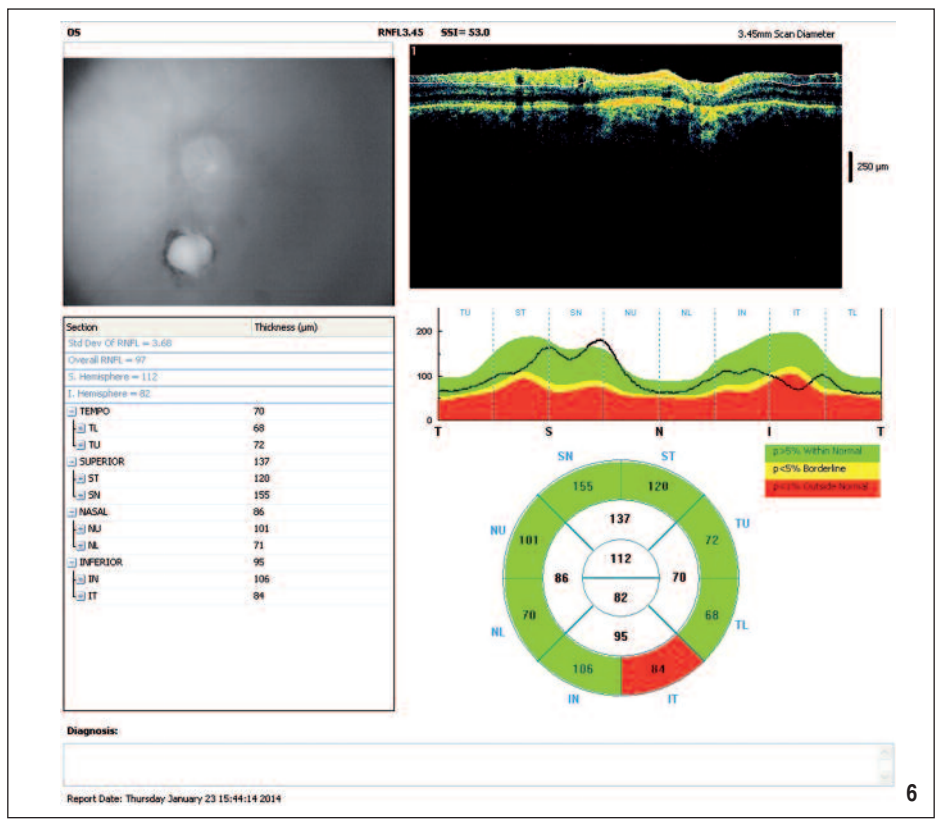


FIGURE 5, 6: OCT retinal nerve fiber layer photographs of both eyes.

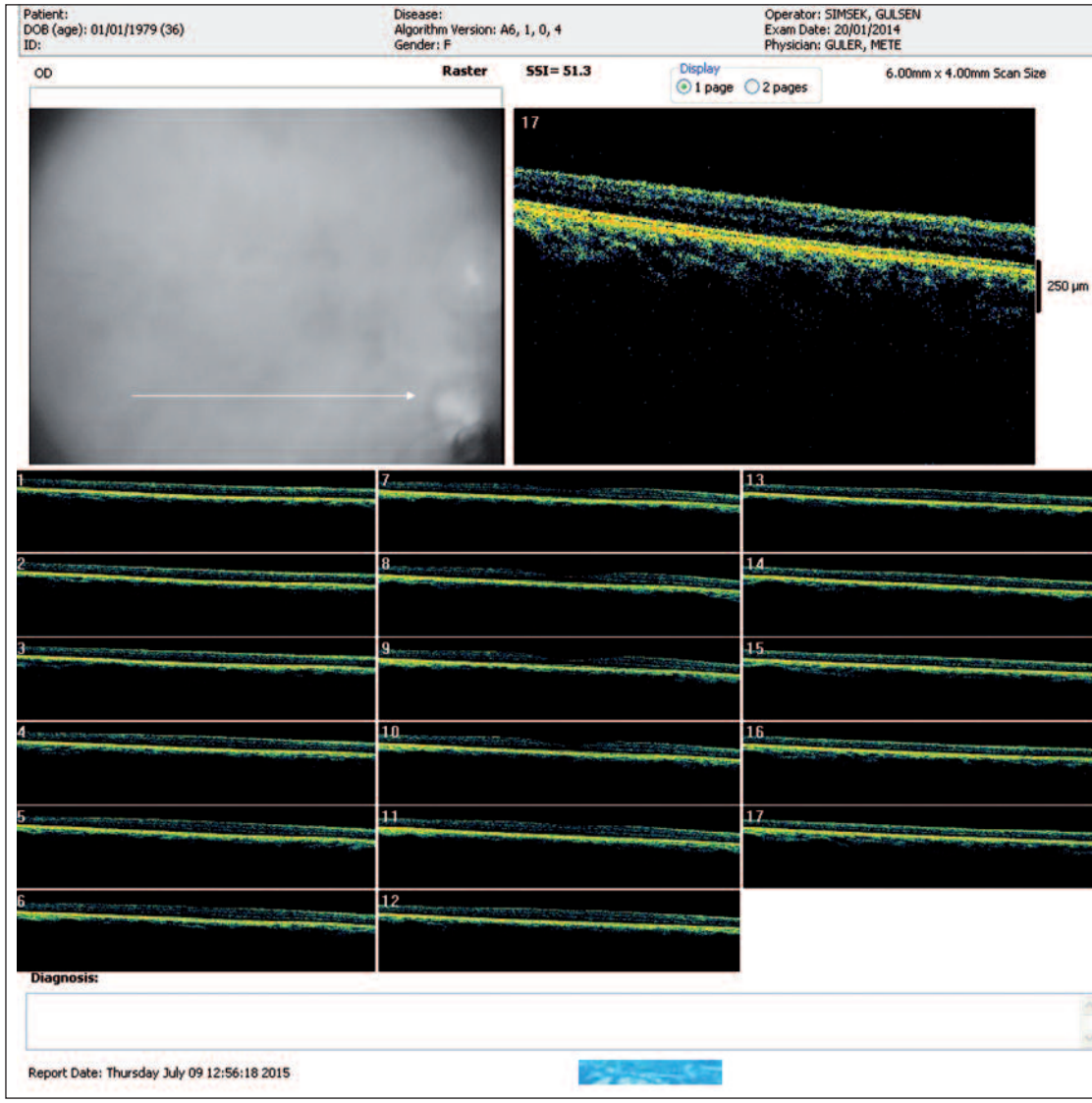


FIGURE 7: OCT raster scans of the right eye.

than chorioretinal coloboma (Figures 3, 4). In optical coherence tomography both infero-temporal retinal nerve fiber layer showed some defects (Figures 5, 6). OCT raster-scans of both eyes were performed (Figures 7, 8). Computerized visual field exam could not be performed due to poor cooperation and exotropia. Extraocular muscles, optic nerve (single optic nerve in each orbit) and retrobulber structures were normal in both eyes under magnetic resonance evaluation (MRI).

DISCUSSION

Ocular coloboma is a congenital, common, and heterogeneous malformation which includes a spectrum of anomalies ranging from iris coloboma to clinical anophthalmos.[5] It results from failure of closure of the fetal fissure. The fetal fissure closes first in the equatorial area then continues to close anteriorly and posteriorly from the equator.[6] Choroidal coloboma is usually an inferiorly and slightly nasally located congenital lesion characterized by absence of the normal retina, retinal pigment epithelium, and choroids. Some patients with many malformations can have severely impaired vision, whereas others have only a cosmetic manifestation, and still others may have no symptoms and may go undiagnosed or may be detected incidentally on routine eye examination. Although

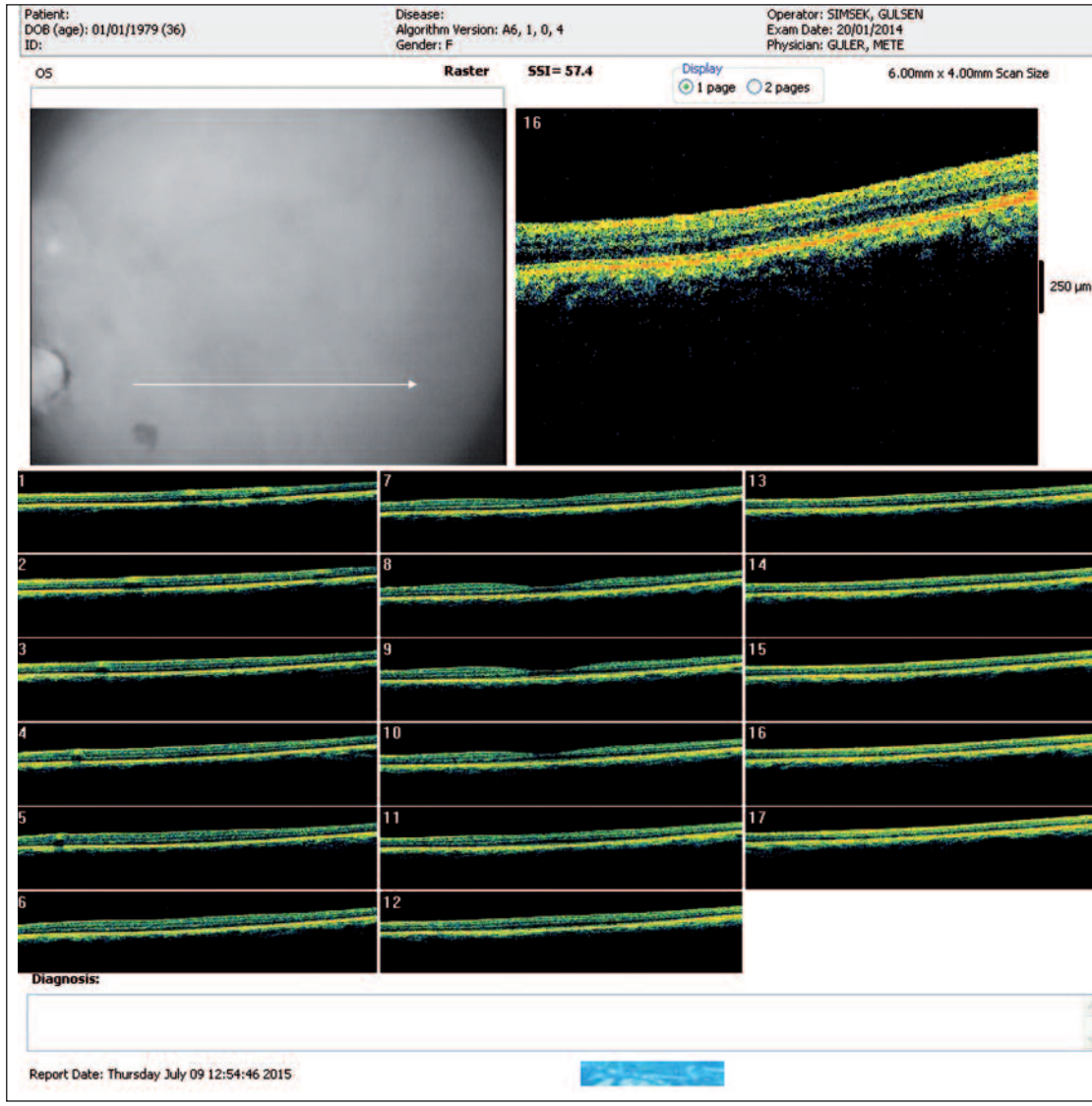


FIGURE 8: OCT raster scans of the left eye.

chorioretinal colobomas may be totally asymptomatic, and may be present early in life, in most cases a hereditary pattern could not be established, as the patient described here in.[7] As an isolated defect, it is usually inherited as an autosomal dominant disorder, although autosomal recessive inheritance also occurs.

True optic disc duplication with two independent retinal vasculatures is rare. In this case, emergence of blood vessels from the center of the colobomatous area and the colobomatous area itself were resembling an accessory optic disc. There was a single optic nerve in each orbit under MRI evaluation. Identification of bridging retinal vessels from the natural optic disc to the colobomatous area which appears as pseudo disc may prevent unnecessary investigations.[6] No management was necessary but the subject was advised to report for visual examination at regular intervals because complications may occur at any age. One should be aware of chorioretinal colobomas may resemble accessory optic disc.

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