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SHORT COMMUNICATION

Case report

Rieger anomaly with bilateral choroidal osteoma: Coincidence or association?

K. ERKILIÇ, A. ÖZKIRIŞ, C. EVEREKLIOGLU, H. DOGAN

Department of Ophthalmology, Erciyes University Medical Faculty, Kayseri - Turkey

PURPOSE. To report a case of Rieger anomaly associated with bilateral choroidal osteoma.
CASE. An 18-year-old woman presented with corectopia, iridocorneal adhesions, and stromal hypoplasia of the iris, and was diagnosed with Rieger anomaly.
RESULTS. Fundus examination revealed bilateral yellowish-orange, placoid degenerative fundus lesions with pseudopod-like edges associated with the areas of retinal pigment epithelium atrophy, indicating choroidal osteoma. Fundus fluorescein angiography showed bilateral early patchy and late diffuse hyperfluorescence. B-scan echography revealed placoid lesion at the posterior ocular coats characterized by localized areas of high ultrasound reflectivity with a corresponding retrobulbar orbital shadowing. This indicated a dense calcium foreign body. In addition, computed tomography of the orbit demonstrated bilateral plate-like thickening with calcification of the choroid that was isodense with the normal skeletal bone.
CONCLUSIONS. Although Rieger anomaly is classically known as a disease of the anterior segment of the eye, choroidal osteoma may be a posterior segment finding of the disease that has not previously been reported. (Eur J Ophthalmol 2003; 13: 496-9)

KEY WORDS. B-scan ultrasonography, Choroidal osteoma, Computed tomography, Fundus fluorescein angiography, Rieger anomaly

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INTRODUCTION

Rieger anomaly and choroidal osteoma are well-defined entities with distinctive clinical and pathologic features. Rieger anomaly is an autosomal dominant disorder resulting in abnormal development of the anterior segment of the eye (1, 2). Choroidal osteoma is an uncommon, benign acquired bony tumor of the peripapillary choroid that occurs predominantly in healthy older juvenile and young adult women (3). Choroidal osteoma occurs between the second and third decades of life and bilateral occurrence is not unusual; it may be a juxtapapillary lesion or the entire disc may be surrounded by the tumor, threatening vision. Although familial cases have occasionally been reported (4), the presence of Rieger anomaly with choroidal osteoma has not previously been reported.

Case report

An 18-year-old woman was admitted to our outpatient clinic with decreased vision. Her ocular examination revealed a visual acuity of 1/10 in the right eye and finger counting from 3 m in the left eye. The hor-

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izontal corneal diameter of the right eye was 10 mm and there were corectopia and iridocorneal adhesions of the iris with stromal hypoplasia (Fig. 1). The horizontal corneal diameter of the left eye was 11 mm and there were strands of peripheral iris to the cornea in front of the Schwalbe line. Intraocular pressures were normal in both eyes. There were no additional changes in the anterior segment.

Ophthalmoscopic examination showed that there were bilateral yellowish-orange, well-defined degenerative fundus lesions associated with disruption and atrophy of the overlying macular retinal pigment epithelium as well as of the sensory retina (Fig. 2, a and b). The surface of the lesion was visibly uneven with depressions and elevations in the left eye. The margins of choroidal osteomas were somewhat irregular in contour, with well-defined scalloped or pseudopod-like edges. The lesions measured about 4 disc diameters in the right eye and 7 disc diameters in the left eye. The lesion was predominantly on the macular region in the right eye whereas the lesion was more extensive in the left eye, with macular and nearly circumpapillary localization. All these fundus findings clearly indicated bilateral choroidal osteoma.

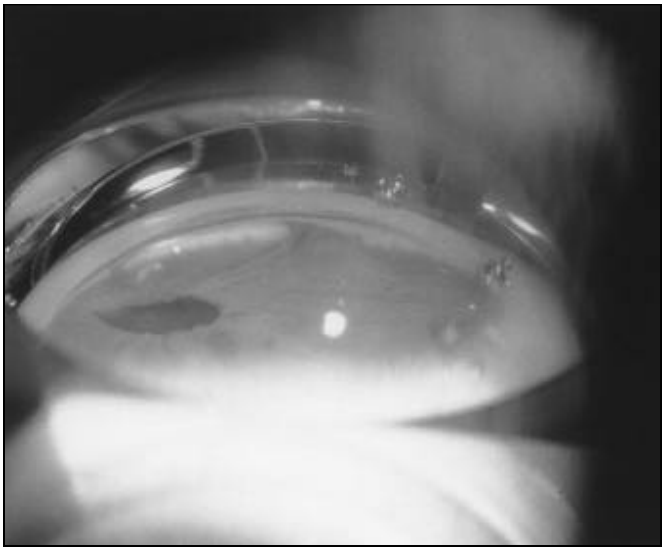

Fig. 1 - Corectopia and anterior chamber angle showing broad leaves of iris stroma adhered to the cornea in the right eye.

Fundus fluorescein angiography of the right (Fig. 3a) and left (Fig. 3b) eyes showed bilateral early patchy and late diffuse hyperfluorescence. There was no choroidal neovascularization in either eye. B-scan ultrasound revealed bilateral high acoustic reflection of the placoid lesions with retrobulbar orbital shadowing, indicating calcification. Computed tomography of the orbit clearly demonstrated bilateral posterior pole lesions with calcification and showed the plate-like thickening of the choroid that was isodense with normal skeletal bone (Fig. 4). The manifestations of Rieger anomaly were prominent in the right eye whereas choroidal osteoma was larger in the left eye.

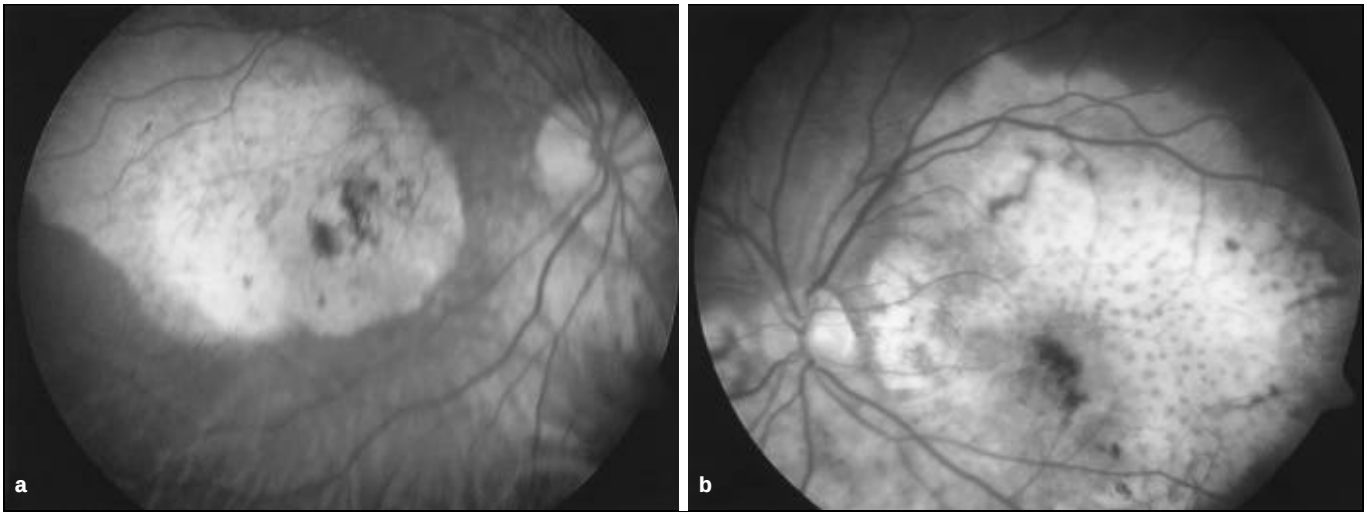

Fig. 2 - Bilateral degenerative fundus lesions associated with the areas of retinal pigment epithelium atrophy and characteristic pseudopod-like edges that measured about 4 disc diameters in the right eye **a)** and 7 disc diameters in the left eye **b)**.

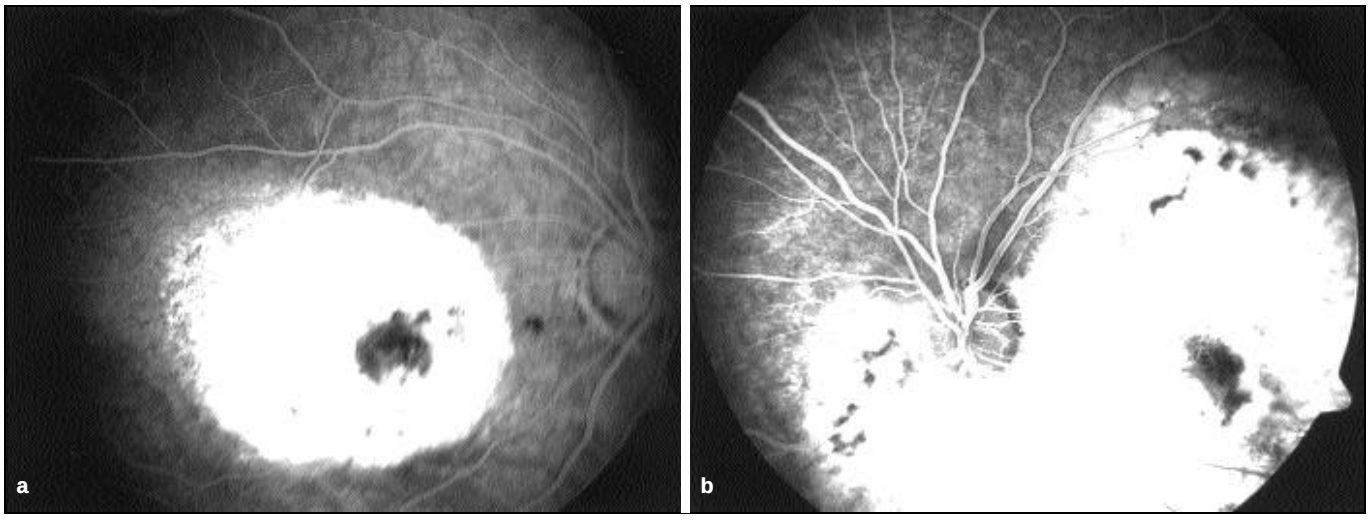

Fig. 3 - Fundus fluorescein angiography of the same patient showed bilateral hyperfluorescence in the right**a)** and left **b)** eyes.

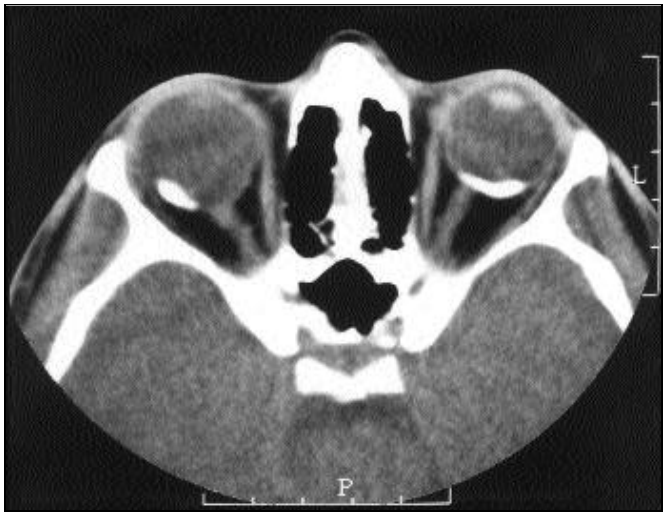

Fig. 4 - Computed tomography of the orbit accentuated the calcification and showed bilateral plate-like thickening of the choroid that was isodense with normal skeletal bone.

DISCUSSION

Rieger anomaly with choroidal osteoma has not previously been reported. Rieger anomaly is a rare condition involving anterior segment dysgenesis and is inherited as an autosomal dominant trait (1). It involves a spectrum of ocular findings consisting of posterior embryotoxon, midperipheral iris adhesions (leukomas) to the cornea, and iris anomalies (iris hypoplasia, pseudopolycoria, corectopia, ectropion uvea) (1, 2). Microcornea and macrocornea may accompany this condition. Although glaucoma develops in about 50% of affected individuals, our patient had normal intraocular pressure in each eye.

Ossification of the ocular coats may be caused by a number of etiologic factors. The bone develops in the tissues owing to altered metabolism as a result of calcium deposition, which acts as a stimulant for osteoblastic activity. Bony tissue may also develop as a result of the activity of displaced osteoblasts, without any systemic or local metabolic disorder. Moreover, metastatic calcification, inflammation, abnormal calcium or phosphorus metabolism, trauma, phthisis bulbi, and longstanding retinal detachment may be the etiology of bone formation within the choroid. On the other hand, although the stimulus for development of choroidal osteomas is not known, these are probably not true choristomas because these lesions do not appear to be congenital in nature. Furthermore, the majority of affected patients are women. Therefore, hormonal or endocrine factors may have a role in the development of these lesions (3, 5). Visual acuity can be substantially impaired in such patients on the basis of degeneration of the overlying retinal pigment epithelium and neurosensory retina. This was the case in our patient and visual acuity was affected much more in the left eye than in the right. Bone formations of the lesions in our patient were confirmed

by ultrasonography and computed tomography. Because the lesions consist of dense bone, echography showed a highly reflective plate-like tissue that shadows the retrobulbar orbital area. On the other hand, we did not observe any relationship between the severity of Rieger anomaly and choroidal osteoma because the findings of Rieger anomaly were prominent in the right eye whereas the dimensions of choroidal osteoma were larger in the left eye.

Although choroidal osteomas have been noted to enlarge in limited fashion during several years of observation, such lesions appear to have no malignant potential (6). Possible explanations for the development of choroidal osteoma in patients with Rieger anomaly are that 1) a coexistence of the two separate diseases may exist or 2) this is a very rare feature of a single disease. Likewise, the chromosomal abnormalities of Rieger anomaly (7) may have affected calcium metabolism, causing development of choroidal osteoma. Future studies with large series may help us to clarify the exact cause of this unique disorder.

In conclusion, this case is the first report of Rieger anomaly associated with bilateral choroidal osteoma. We did not demonstrate any causal link between the two entities and, therefore, coincidence seems to be the most likely explanation.

Reprint requests to:
Cem Evereklioglu, MD
Sivas Cad. Cebeci Apt. A-Blok, 175/15
TR-38020, Kayseri, Turkey
everekliogl u@hotmail.com

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