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OPHTHALMOLOGY
Case Report

Combined Hamartoma of Retina and Retinal Pigment Epithelium calcified as an incidental finding: Case report

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Abstract

Introduction: Combined hamartoma of the retina and retinal pigment epithelium (CHR- RPE) is an uncommon and benign tumor, typically single and unilateral, which may compromise the optic nerve, macula and less frequently the periphery.

Case Report: A 59-year-old woman, with a history of strabismus surgery in childhood. She refers decreased visual acuity unilaterally in the right eye, without other symptoms. She had a visual acuity of light perception in the right eye, highlighting the fundoscopy that showed a yellow-gray mass, elevated above the optic disc and peripapillary retina, extending to macula. The study was completed with fluorescein angiography, ocular ultrasound, and optical coherence tomography (OCT), establishing the diagnosis of CHR-RPE. Patient is currently in control.

Discussion: This lesion is characterized by proliferation of retinal pigment epithelium and glial tissue, causing great papillary and retinal distortion, vascular tortuosity, hyper pigmentation, and sometimes epiretinal membranes. Its course is steady and its growth is exceptional. Most do not have systemic manifestations. There are reported cases with neurofibromatosis type I and II, although this association is not well established yet.

Conclusion: The diagnosis is not easy and in some cases requires excisional biopsy. Although it is benign must make an adequate differential diagnosis and monitoring.

Keywords: *Intraoculars tumors; Combined hamartoma; Retina; Pigment epithelium*

Abbreviations: (CHR- RPE): Combined hamartoma of the retina and retinal pigment epithelium;

Introduction

Combined hamartoma of the retina and retinal pigment epithelium (CHR- RPE) is an uncommon and benign ocular tumor [1]. Typically single and unilateral, which may compromise the optic disc, macula and less frequently the periphery. This congenital lesion consists of glial cells, vascular tissue, and sheets of pigment epithelial cells. It is commonly found in healthy young children; however it has been described some cases associated to systemic diseases [2]. We report a case of a CHR- RPE.

Clinical Case

A 59-year- old woman, with a history of strabismus surgery in childhood, refers a decreased visual acuity in the right eye, without other symptoms. Her visual acuity was light perception OD and 20/20 OS. Fundus examination of the right eye showed a yellow-gray mass, slightly elevated above the optic disc and peripapillary retina, extending to macula and hyper pigmented areas around the lesion also tortuous vessels (Figure 1) the left eye fundoscopy was normal. The fluorescein angiography showed a hyper fluorescent lesion, without leakage (Figure 2), B-scan ultrasound showed a mildly elevated lesion above the optic disc level with calcified nodular areas. (Figure 3) OCT demonstrated an elevated lesion, with a loss of the normal architecture of the optic nerve and gradual transition from this

lesion to the normal adjacent retinal layers, the retinal pigment epithelium is intact. (Figure 4), in the image of the macula demonstrates loss of the foveal depression and the presence of a hyper-reflecting nodule (Figure 5). With all this findings the diagnosis of CHR-RPE was established. Patient is currently in control.

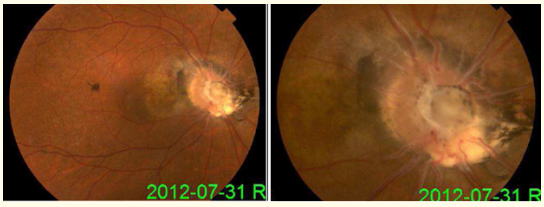

Figure 1: *The fluorescein angiographya tumoral hyperfluorescent lesion.*

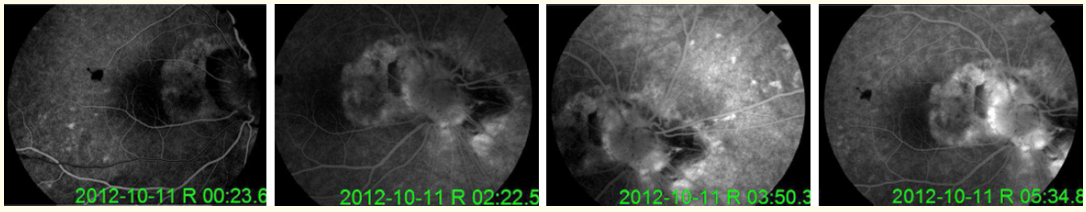

Figure 2: *B-scan ultrasound showed a mildly elevated lesion.*

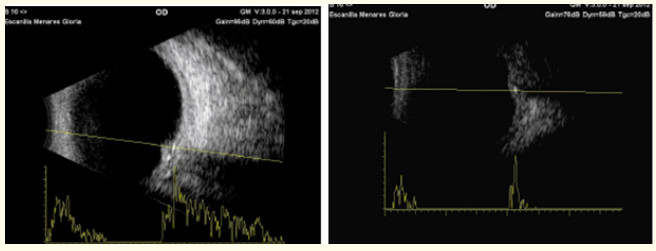

Figure 3: *OCT demonstrated an elevated lesion.*

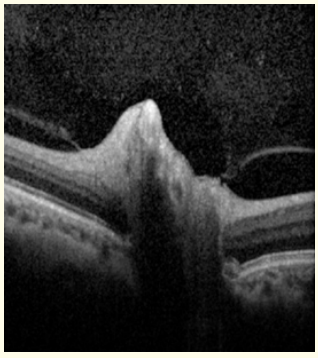

Figure 4: *Loss of the foveal depression and the presence of an epiretinal membrane.*

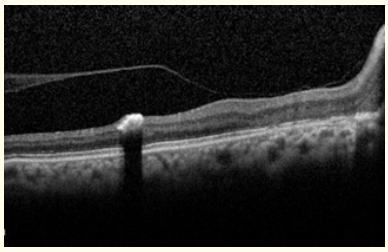

Figure 5: *Diagnosis of CHR-RPE was established.*

Discussion

The CHR-RPE is an uncommon ocular tumor. It was first described by Gass in 1973, who reported 7 cases emphasizing the difficulty of diagnosis as it could simulate malignant tumors as choroidal melanoma and retinoblastoma [1]. Clinically, it is characterized by a slightly elevated lesion usually pigmented and a central portion with gliosis and tortuous vessels [1]. The tumor usually presents as a unilateral solitary lesion close to the optic disc, or in the juxtapapillary or macular area, although it may occur peripherally as well (5%) [3]. its course is steady and its growth is exceptional [2]. Histopathologically there is proliferation of retinal pigment epithelium and glial tissue, altering the normal retinal architecture [1,2] causing great papillary and macular distortion, vascular tortuosity, hyperpigmentation, and epiretinal membranes [3]. It has also been described an association with preretinal and coroidal neovascularization [4,5]. In the case we found a unilateral lesion in the right eye, slightly elevated above the optic disc and peripapillary retina that extended to macula, with hyperpigmented areas around the lesion. The CHR-RPE has been associated with some systemic diseases, as neurofibromatosis type II and less commonly type I, Gorlin Goltz syndrome, Poland anomaly, branchio-oculo-facial syndrome, and juvenile nasopharyngeal angiofibroma. These associations are not well established yet [2-6]. In the case we report, there were not systemic manifestations. The CHR-RPE is usually found in young children, often with decreased visual acuity (less than 20/200 in 47% of cases), leukocoria or strabismus [2]. Schachat., *et al*. reported a mean age of diagnosis of 15 years old [7]. In the case we report, the patient was an adult but refers a long time with a unilateral decrease in visual acuity and a history of strabismus in childhood. An appropriate diagnosis is imperative to prevent unnecessary enucleation [8]. The fluorescein angiography, ultrasound examination and OCT are useful in establishing the diagnosis and also to exclude malignant lesions and to perform an accurate follow-up [9,10]. The fluorescein angiography commonly shows early hyperfluorescence throughout the lesion [9]. Ocular ultrasound may show a slightly elevated solid lesion and could be associated to calcification areas. In this case we found a lesion with this characteristic. The OCT typically shows a hypereflective lesion and hyporeflective shadowing the underlying tissue. Is also useful to detect epiretinal membrane, macular traction and macular edema [96,10]. Pars Plana vitrectomy and epiretinal membrane peeling has been proposed as a surgical treatment that could improve visual acuity, however in most cases this is not possible to predict since CHR- RPE presents at a young age and amblyopia could have been developed [11,12]. In most cases the appropriate management is to exclude malignant lesions and perform an accurate follow-up.

Conclusion

The diagnosis of CHR- RPE could be difficult and in some cases requires excisional biopsy. Although it is a benign tumor, it is important an adequate differential diagnosis and to follow–up these patients in order to detect malignant tumors, as retinoblastoma in children.

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