INTRODUCTION

Combined Hamartomas of Retina and Retinal Pigment Epithelium (CHRRPE) are rare benign tumors characterized by the proliferation of retinal pigment epithelium (RPE) and gial tissue, generating a large papillary and retinal distortion. A single unilateral over- elevated hyper-pigmented lesion is observed, accompanied by vascular tortuosity and epiretinal membrane (ERM).

OBJECTIVE

To show the long-term evolution of two cases of Combined Hamartoma of Retina and Retinal Pigment Epithelium (CHRRPE), one managed by vitrectomy, and other managed conservatively.

METHODS

A 46-year-old male and a 35-year-old female, diagnosed as having a combined hamartoma of the retina and retinal pigment epithelium, were observed for eleven and four years, respectively. The first case was treated with pars plana vitrectomy due to large associated epiretinal membrane (ERM), and the other patient declined surgery.

RESULTS

PATIENT 1: AT FIRST VISIT. Best corrected visual acuity (BCVA) in RE was 0.1 (decimal notation), and in left eye (LE), 1.0. Anterior segment examination revealed a bilateral nuclear sclerosis with no further abnormalities. Fundoscopy revealed a hyperpigmented and elevated peripapillary lesion, with macular involvement and a huge epiretinal membrane over macular and peripheral region. Tortuous and telangiectatic vessels over the lesion were appreciated. Fluorescein angiography (FA) showed hypofluorescence and telangiectatic vessels with diffusion of contrast in the early frames, and hyperfluorescence in late phases, without signs of neither choroidal nor retinal neovascularization.

Considering the diagnosis of combined hamartoma of the retina and retinal pigment epithelium, the patient was referred to the Internal Medicine department to rule out any associated diseases. A complete systemic examination was performed, including Magnetic Resonance and contrast CT Scan in order to detect the presence of any neurofibroma. According to the diagnostic criteria, neurofibromatosis was excluded.

Due to the important VA impairment and the presence of large ERM, we decided to perform 20-gauge three-port pars plana vitrectomy (PPV) with membrane peeling.

Vitreoretinal surgery resulted in an anatomical improvement and VA stabilization at six months postoperatively. Complete ocular examination was performed every six months, showing the lesion to be unchanged. One year after vitrectomy a cataract developed, so the patient underwent phacoemulsification with intracapsular lens implantation.

FOUR YEARS LATER he complained of visual loss, presenting severe macular distortion, and, consequently, decrease in visual acuity in his right eye, and examination revealed a BCVA in RE of hand movement at 1 meter. Fundoscopy revealed an epiretinal membrane occupying the lesion and telangiectatic vessels with diffusion of contrast in late phases, similar to the previous case. OCT showed the presence of a continuous line of hyperreflectivity attached to the inner retinal surface corresponding to an ERM, and a hyperreflective space corresponding to a neurosensory retinal detachment. OCT also revealed the presence of intraretinal cysts and a feudal defect compatible with a macular pseudohole.

The patient declined surgery, therefore a conservative approach with a rigorous follow-up was performed. Four years later the lesion remained unchanged with a visual acuity of 0.3 in her LE.

CONCLUSION: Vitreoretinal surgery for CHRRPE can improve retinal architecture and visual acuity, but in the long-term functional recuperation is frequently not possible, with similar visual outcomes to those managed conservatively.

REFERENCES