INTRODUCTION

Combined hamartoma of the retina and retinal pigment epithelium are extremely rare benign tumors, which are characterized by retinal pigment epithelium (RPE) and glial tissue proliferation, provoking severe peripapillary and retinal distortion. There are no established managements for combined hamartoma of the retina and retinal pigment epithelium.

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 CHRRPE, were observed during four years. One of them underwent pars plana vitrectomy (PPV) due to an associated epiretinal membrane (ERM), and the other patient declined surgery. Full clinical examination, including Best Corrected Visual Acuity (BCVA), funduscopy examination, Fluorescein Angiography (FA), and Optical Coherence Tomography (OCT) was performed.

RESULTS

PATIENT 1 presented with visual acuity (VA) loss in the right eye (RE). BCVA in RE was 0.1 (decimal notation), and in left eye (LE), 1.0. Anterior segment examination revealed a bilateral nuclear cataract with no further abnormalities. Funduscopy revealed a hyperpigmented and elevated peripapillary lesion, with macular involvement and a huge ERM over macular and perimacular region. Tortuous and telangiectatic vessels over the lesion were appreciated. 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 PPV with membrane peeling. Vitreoretinal surgery resulted in an anatomic improvement and VA stabilization at six months postoperatively. Complete oculic examination was performed every six months, showing the lesion to be unchanged.

However, FOUR YEARS AFTER PPV, the patient presented complaining of decreased vision in his right eye, and examination revealed a BCVA in RE of hand movement at 1 meter. Fundusoscopic examination and OCT (Cirrus SD, Carl Zeiss, Germany) showed the lesion to remain unchanged. OCT also demonstrated severe macular distortion with areas of significant increase in retinal thickness with intraretinal cysts, associated to severe RPE atrophy, all these contributing to poor visual acuity.

PATIENT 2 was referred to our department complaining of progressive loss of vision in her LE. BCVA in RE was 1.0 (decimal notation), and in LE 0.4. Anterior segment examination revealed no abnormalities. Funduscopy showed a hyperpigmented and elevated macular lesion, with an associated epiretinal membrane. FA showed hypofluorescence of 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 neurosensorial retinal detachment. OCT also revealed the presence of intraretinal cysts and a foveal defect compatible with a macular pseudohole.

The patient also showed melanocytic lesions in several regions of the body which remained us to "Café au lait" spots. She was referred to the Dermatology department in order to discard neurofibromatosis. However, the final diagnosis was solar lentigo.

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 the long-term functional recuperation is frequently not possible, with similar visual outcomes to those managed conservatively.

REFERENCES