ABSTRACT

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

Case Reports
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 four years. One of them was treated with pars plana vitrectomy due to large associated epiretinal membrane (ERM), and the other patient declined surgery. Initially, the evolution was satisfactory in the patient who underwent vitreoretinal surgery, with anatomic improvement and visual acuity stabilization. However, four years later, he complained of visual loss, presenting severe macular distortion, and, consequently, decrease in visual acuity. The patient who preferred a conservative management, showed no funduscopic changes during the follow-up.

Conclusion
Vitreoretinal surgery for combined hamartoma of the retina and retinal pigment epithelium may improve retinal architecture and visual acuity, but in the long-term functional recovery is frequently not possible, leading to a decrease in VA.

We can conclude that the evolution of this pathology can be similar in both cases treated with vitreoretinal surgery and in cases without surgery. Therefore, conservative management may be adopted. A larger number of cases are required to be studied to confirm our clinical observations.

Key words: Combined Hamartomas of Retina; Retinal Pigment Epithelium, vitrectomy.
Case Reports

Case 1

A 46-year-old Spanish man with history of a cranioencephalic trauma two years before presentation, who presented with visual acuity (VA) loss in the right eye (RE). Best corrected visual acuity (BCVA) in RE was 0.1 (decimal notation), and 1.0 in the left eye (LE). Anterior segment examination revealed a bilateral nuclear sclerosis with no further abnormalities. Fundoscopy revealed a hyperpigmentated and elevated peripapillary lesion, with macular involvement and a huge epiretinal membrane over macular and perimacular region. Tortuous and telangiectasic vessels over the lesion were noted (Figure 1).

Fluorescein angiography (FA) showed hypofluorescence and telangiectasic vessels with diffusion of contrast in the early frames, and hyperfluorescence in late phases, without signs of neither choroidal nor retinal neovascularization (Figure 2).

Considering the diagnosis of combined hamartoma of the retina and retinal pigment epithelium, the patient was forwarded 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 identify the presence of any neurofibroma. Based upon 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 anatomic improvement and VA stabilization at six months postoperatively. Complete ocular examination was performed every six months, showing the lesion to remain unchanged.

However, four years after PPV, the patient presented complaining of decreased vision in his right eye. Examination revealed a BCVA on RE of hand motion at 1 meter. Fundusscopic examination (Figure 3) and Optical Coherence Tomography (OCT) (Cirrus SD, Carl Zeiss, Germany) (Figure 4) showed the lesion to remain unchanged. OCT also demonstrated severe macular distorsion with areas of significant increase in retinal thickness with intraretinal cysts, associated to severe RPE atrophy, all contributing to poor visual acuity.

Case 2

A 35-year-old Spanish woman, referred to our department complaining of progressive loss of vision in her LE. BCVA in RE was 1.0 (decimal notation), and 0.4 in the LE. Anterior segment examination revealed no abnormalities. Fundoscopy showed a hyperpigmentated and elevated macular lesion, with an associated epiretinal membrane (Figure 5). Fluorescein angiography (FA) showed hypofluorescence and telangiectasic vessels with diffusion of contrast in the early frames, and hyperfluorescence in late phases, without signs of neither choroidal nor retinal neovascularization (Figure 2).

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angiography showed hypofluorescence of the lesion and telangiectatic vessels with diffusion of contrast in late phases, similar to the previous case (Figure 6).

OCT showed the presence of a continuous line of hyporeflectivity attached to the inner retinal surface corresponding to an ERM, and a hyporeflective space corresponding to a neurosensorial retinal detachment. OCT also revealed the presence of intraretinal cysts and a foveal defect compatible with macular pseudohole (Figure 7).

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 (Figure 8).

Discussion

The progression of combined hamartoma of the retina and retinal pigment epithelium is used to be stationary.

This is why most cases require no treatment. However, VA decrease related to choroidal neovascularization, vitreous hemorrhage, macular edema, macular hole or retinal detachment formation are described in the literature.1,4–5

There have been several management options described such as photodynamic therapy with Verteporfin6 (for cases with vascular leakage), a combination therapy consisting of vitrectomy, laser photoacoagulation and intravitreal triamcinolone3, and vitrectomy associated to ERM peeling2, all of them with good surgical outcomes and a low rate of intra and postoperative complications. The recurrence of the ERM is possible, but most authors consider that VA may improve despite this recurrence.8

We report two cases of combined hamartoma of the retina and retinal pigment epithelium with different management approaches (PPV with ERM peeling in one case and conservative treatment in the other case). Both cases showed a similar evolution in the long term, after four years of follow-up.

Conclusion

Vitreoretinal surgery for combined hamartomas of retina and retinal pigment epithelium may improve retinal architecture and visual acuity in the short term.

However, functional outcomes in the long term can be identical to those managed conservatively. This is why we propose a conservative management approach of combined hamartoma of the retina and retinal pigment epithelium, especially for those hamartomas with macular involvement and severe retinal distortion.

References