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Case Report

Epiretinal membrane associated with combined hamartoma of retina pigment epithelium: Surgical outcome and histopathological features

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ABSTRACT

Background: Combined hamartoma of the retina and retinal pigment epithelium (CHR-RPE) represents a rare, benign congenital anomaly typically presenting as slightly elevated, variably pigmented lesions. This report details the surgical outcome and histopathological features of an epiretinal membrane associated with CHR-RPE.

Materials and Methods: A 29-year-old woman with a three-year history of gradual visual decline in her right eye underwent a comprehensive ophthalmologic examination, including spectral domain optical coherence tomography (SD-OCT), which revealed characteristic features of CHR-RPE and an overlying epiretinal membrane. She subsequently elected to undergo pars plana vitrectomy with membrane peeling.

Results: Intraoperative findings confirmed the presence of histological elements consistent with the internal limiting membrane, fibrillar collagen, vitreous, and cells positive for glial fibrillary acidic protein (GFAP), indicative of glial cells. Postoperatively, there was a noticeable improvement in the retinal architecture; however, visual acuity improvement was minimal. Histopathological analysis of the excised membrane provided insight into the lesion's cellular composition.

Conclusion: This case underscores the complex nature of CHR-RPE associated with epiretinal membranes and the challenges in achieving significant visual acuity improvement despite successful surgical intervention. It highlights the importance of early detection and a personalized approach to management. Future research should focus on optimizing treatment strategies and improving understanding of the prognosis for similar cases.

1. Introduction

In the evolving landscape of retinal surgery and pathology, the intersection of advanced imaging techniques and surgical interventions has catalyzed significant improvements in the diagnosis and management of complex retinal diseases. The case report titled "Epiretinal Membrane Associated with Combined Hamartoma of Retina Pigment Epithelium: Surgical Outcome and Histopathological Features" exemplifies this progress through the meticulous examination of a unique clinical scenario. The focal point of this report is a 29-year-old female patient who presented with a gradual, painless decline in visual acuity in her right eye, a condition that lacked any significant medical history which could predispose her to ophthalmic issues.[1]

Upon initial evaluation, the clinical and imaging findings pointed towards a combined hamartoma involving the retina and retinal pigment epithelium (RPE), an uncommon and benign congenital malformation. Characterized by a mixture of tissues including glial cells, blood vessels, and pigmented epithelial cells, combined hamartomas of the retina and RPE manifest as slightly raised, pigmented lesions, typically occurring unilaterally and without associated systemic conditions. The complexity

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of this case was further amplified by the presence of an epiretinal membrane, a fibrocellular layer that can exert traction on the retina, potentially leading to visual impairment.[2]

The intricacies of this condition were unraveled through spectral domain optical coherence tomography (SD-OCT), which provided detailed imagery of the disorganized retinal architecture and the hyperreflective thickening indicative of the hamartoma and the overlying epiretinal membrane. The decision to proceed with a pars plana vitrectomy, a standard surgical approach for the removal of the vitreous humor along with the epiretinal membrane, was grounded in the hope of restoring retinal anatomy and, subsequently, visual function. This surgical endeavor not only offered the opportunity for anatomical correction but also for the acquisition of tissue for histopathological analysis, an invaluable asset for understanding the cellular makeup of these complex lesions.[3]

Histopathological examination shed light on the nature of the epiretinal membrane, revealing a composition of basement membrane-like material, fibrillar collagen, and cells positive for glial fibrillary acidic protein (GFAP), indicating the presence of glial cells. These findings underscore the heterogeneity of the tissue components involved in the formation of epiretinal membranes associated with combined hamartomas, illuminating the pathological processes underpinning this condition.[4]

While the surgical intervention led to an improvement in the structural integrity of the retina, it underscored a crucial reality in the management of such conditions: anatomical success does not always translate into functional recovery. The patient's visual acuity remained largely unchanged post-surgery, a reminder of the challenges that persist in treating retinal diseases where the damage to visual pathways may not be fully reversible.[5]

2. Case Presentation

We present an exceptional case involving a 29-year-old female with no noteworthy medical, surgical, or ocular background, who approached our clinic with complaints of a three-year progression of visual impairment in her right eye. The patient did not report additional visual disturbances and had a non-significant social history. Upon clinical evaluation, her visual acuity was measured at 20/200 in the affected eye and 20/20 in the unaffected eye, with normal intraocular pressures of 15 mmHg in both eyes. No afferent pupillary defect was detected, and extraocular muscle movements were deemed normal. The anterior segment examination was normal for both eyes.

A dilated fundus examination of the right eye revealed an unevenly defined, pigmented, slightly elevated retinal lesion in the papillomacular region, covered centrally by a dense, gray, contracted epiretinal fibrous membrane. This membrane seemed to warp and draw the adjacent retina and retinal blood vessels toward the center of the lesion. The fundus examination of the left eye showed no abnormalities.

Spectral-domain optical coherence tomography (SD-OCT) for the right eye depicted a disorganization across multiple retinal layers with an overlaying epiretinal membrane, resulting in folded, disorganized hyperreflective retinal thickening. This led to hyporeflective shadowing of the tissue underneath, indicating a combined hamartoma of the retina and retinal pigment epithelium (RPE) accompanied by an epiretinal membrane.

After a thorough discussion of the diagnosis and potential outcomes, the patient chose to undergo pars plana vitrectomy with membrane peeling, aiming to improve her visual acuity by addressing the epiretinal membrane. The surgery employed a conventional three-port vitrectomy approach, initiating with a core vitrectomy, followed by the induction of a posterior vitreous detachment and careful peeling of the epiretinal membrane, facilitated by brilliant blue dye and ILM forceps. The excised epiretinal membrane underwent histopathological examination.

This case illuminates the surgical and diagnostic complexities encountered with combined hamartomas of the retina and RPE, particularly when an epiretinal membrane complicates the condition. Despite pars plana vitrectomy with membrane peeling potentially enhancing anatomical conditions, it may not invariably lead to substantial functional visual improvements. This accentuates the necessity of managing patient expectations regarding the potential visual outcomes post-surgery. The imperative for further investigation into the pathophysiology and optimal treatment strategies for such intricate cases remains, to advance patient care and outcomes.

Optical coherence tomography (OCT) revealed the presence of folded, disorganized hyperreflective retinal thickening that resulted in hyporeflective shadowing of the underlying tissue, accompanied by an overlying epiretinal membrane (Figure 1 A&B). The patient received a diagnosis of combined hamartoma of the retina and retinal pigment epithelium (RPE) in the right eye, with an assessment to exclude amblyopia in the same eye. The patient opted for pars plana vitrectomy with membrane peeling.

Surgical Technique: A standard three-port vitrectomy was executed. The procedure commenced with a core vitrectomy followed by the induction of a posterior hyaloid detachment, which adhered firmly. A thick epiretinal membrane was removed and subsequently submitted for histopathological examination. At the conclusion of the surgery, a thorough inspection of the retinal periphery was conducted to identify any potential peripheral retinal tears, which were found to be absent.

Histopathological Examination: A membrane specimen measuring 2 x 1 mm, stained with eosin from the right eye, was processed in its entirety. The specimen comprised a cross-section through basement membrane-like material,

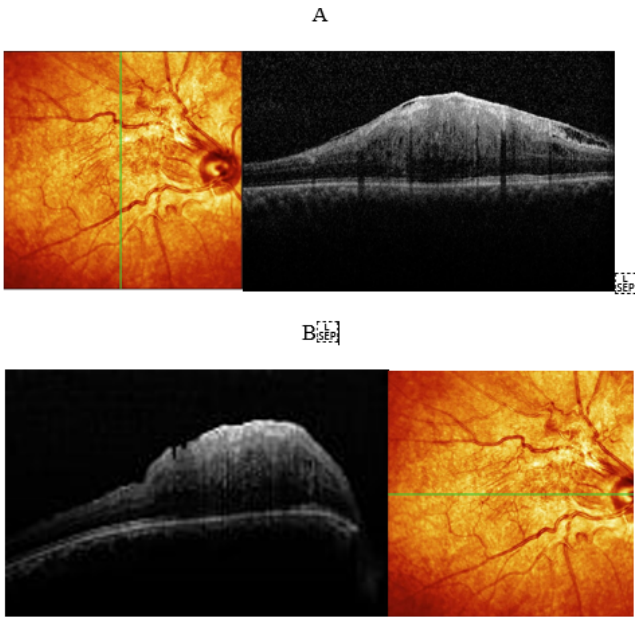

Figure 1: Spectral-domain optical coherence tomography of the lesion. There is disorganization of multiple retinal layers with an overlying epiretinal membrane

consistent with the internal limiting membrane, fibrillar collagen, and vitreous. The diagnostic outcome confirmed the presence of an internal limiting membrane and a glial membrane embedded within a fibrillary matrix.

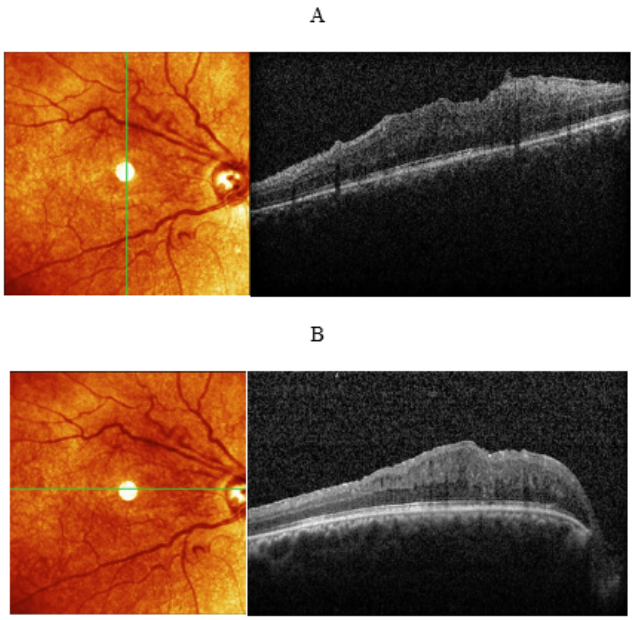

Figure 2: Oct through the macula of the right eye A & B showed resolved epiretinal membrane post surgery with disorginzed retinal layers

The surgery result in improved retinal architecture and no significant improvement in visual acuity 6 month follow up showed no improvement of vision VA 20/200, with no recurrence of epiretinal membrane. (Figure 2 A & B)

3. Discussion

Combined hamartoma of the retina and retinal pigment epithelium (RPE) typically manifests as a slightly elevated lesion, varying in pigmentation on its surface, and featuring a core of dilated, tortuous vessels and gliosis.[6] These lesions mainly occur juxtapapillary or in the macular region, with a minority (approximately 5%) appearing in peripheral locations. The importance of accurately distinguishing these lesions lies in their potential misidentification, which can lead to confusion with intraocular malignancies such as malignant melanoma or retinoblastoma, emphasizing the need for precise diagnosis to prevent unnecessary enucleation.[7]

In-depth analysis of optical coherence tomography (OCT) findings in Combined Hamartoma of the Retina and Retinal Pigment Epithelium (CHRRPE) has identified unique features such as multiple small hyperreflective triangular alterations at the lesion edges, termed the "shark-teeth" sign, along with the disruption and morphological changes in all retinal plexuses. In the case presented, the late onset of visual impairment, attributed to the recent contraction of the epiretinal membrane, was notable.[8] OCT imaging showed a substantial epiretinal membrane with areas of vitreoretinal traction exacerbating the thickened retinal mass, prompting surgical intervention through pars plana vitrectomy for membrane removal. Despite anatomical improvements post-surgery, there was no notable enhancement in visual acuity.[9]

Postoperative reduction in vascular tortuosity and retinal vascular leakage could lead to functional and anatomical improvements. Timely removal of epiretinal membranes in patients with combined retinal and RPE hamartomas may be beneficial. Especially in the pediatric demographic, improvement in retinal structure and visual acuity is achievable with pars plana vitrectomy and membrane peeling, potentially augmented by autologous plasmin enzyme for epiretinal membranes associated with combined hamartomas.[10–14]

Histopathological analysis of an enucleated globe, initially diagnosed as juxtapapillary malignant melanoma, revealed a peripapillary mass composed of disorganized retinal tissue intermixed with vascular, glial elements, and proliferating retinal pigment epithelium tubules, consistent with the internal limiting membrane and a glial membrane embedded in a fibrillary matrix.[15]

The recent decrease in vision in this case suggests the possibility of an acquired hamartoma; however, the congenital nature of the lesion, indicated by the poor visual outcome, suggests it was present from birth but only

recently manifested vision loss. In a series of 20 patients with epiretinal membrane, one case secondary to combined hamartoma showed improved visual acuity post-vitrectomy, likely due to the sparing of the central macula and the extension of the epiretinal membrane over the macula. While favorable surgical outcomes in combined hamartoma cases have been reported, the most extensive surgical series to date reveals a tendency for ongoing glial proliferation and recurrent epiretinal membrane formation post-surgery, with recurrence observed in 4 out of 11 patients. [16,17]

In conclusion, the patient described herein exhibited classic clinical characteristics of combined RPE hamartoma, with additional findings of epiretinal membrane contraction causing displacement of adjacent retina and retinal blood vessels towards the tumor center, alongside recent vision deterioration. Despite undergoing surgical removal of the epiretinal membrane, there was no improvement in visual acuity, suggesting a link to the recent vision loss. [18]

4. Conclusion

The study presented a rare case of epiretinal membrane associated with combined hamartoma of the retina and retinal pigment epithelium, meticulously detailing the surgical intervention and histopathological findings. Despite the surgical improvement in retinal architecture, the patient's visual acuity saw minimal enhancement, highlighting the complex nature of such cases. This underscores the need for early detection and a personalized approach to management, emphasizing that structural rectification does not always correlate with functional recovery. Further research is essential to explore optimal treatment strategies and to better understand the prognosis for similar cases.

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6. Conflict of Interest

None.

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