

# *Diagnostic and Therapeutic Challenges* \_\_\_\_\_ Edited by H. Richard McDonald

Drs. Steven M. Cohen, Elias Reichel, Jorge Guillermo Arroyo, and Brandon J. Lujan

This case was submitted by Dr. Steven M. Cohen, of Retina Vitreous Associates of Florida, Clearwater, Florida, and commented by Dr. Elias Reichel, Boston, Massachusetts, Dr. Jorge Guillermo Arroyo, Boston, Massachusetts, and Dr. Brandon J. Lujan, San Francisco, California.

## Case Report

A 42-year-old man has had darkening central vision in the left eye for 6 months. The patient reports that the left eye has always been more nearsighted than the right eye but was correctable to 20/20. Ten years ago, he noticed a mild loss of vision in the left eye, and 5 years ago, he noticed distortion of the vision in the left eye.

Medical records are available from 4 years ago until last month from his optometrist. His best-corrected vision has fluctuated between 20/30 and 20/70 in the left eye. His most recent refraction was + 1.50 × 0.50 × 118 20/20 in the right eye and + 1.25 – 0.50 × 076 20/30–2 in the left eye from 6 weeks before presentation.

The patient recalled being involved in an automobile accident 18 years ago when he went off the road in the rain, crashed his car, and smashed his face into the steering wheel. He had no loss of consciousness. He was evaluated at the emergency room and was told his left eye, with a swollen, bruised eyelid, was undamaged. His vision from his left eye was blurred for 2 weeks after the accident and then returned to its usual slightly weak level.

His medical history, family history, and social history are benign.

## Examination

The patient's right eye was 20/20 and normal. His left eye had best-corrected visual acuity of 20/60. Intraocular pressures and color vision were normal. Anterior segment examination was normal with a clear lens. Fundus examination in the left eye showed no vitreous cells. Centered on the superotemporal distorted retinal vessels above the macula is a well-circumscribed tractional retinal detachment with an overlying epiretinal membrane and an underlying

circular area of retinal pigment epithelial (RPE) hyperpigmentation (Figure 1).

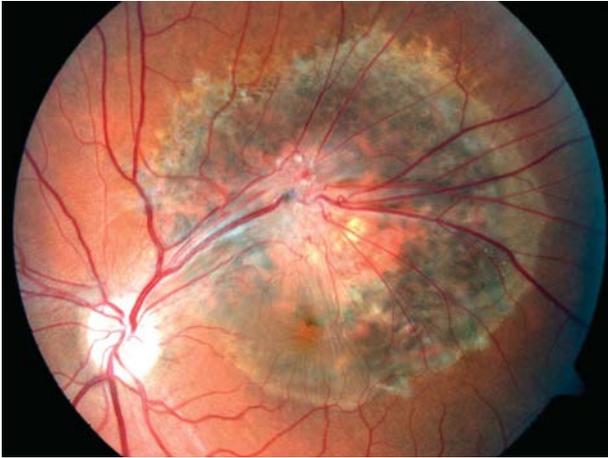
The fluorescein angiogram shows hypofluorescence in the center of the lesion (Figure 2) with late leakage at the edge of the lesion and from the retinal vessels under traction in the center of the lesion (Figure 3). Optical coherence tomography (OCT) shows a staphyloma with an overlying tractional retinal detachment that involves the fovea (Figure 4). B-scan ultrasound shows a shallow staphyloma (Figure 5).

This case is presented for discussion of diagnosis and management.

## Dr. Elias Reichel (Boston, Massachusetts): \_\_\_\_\_

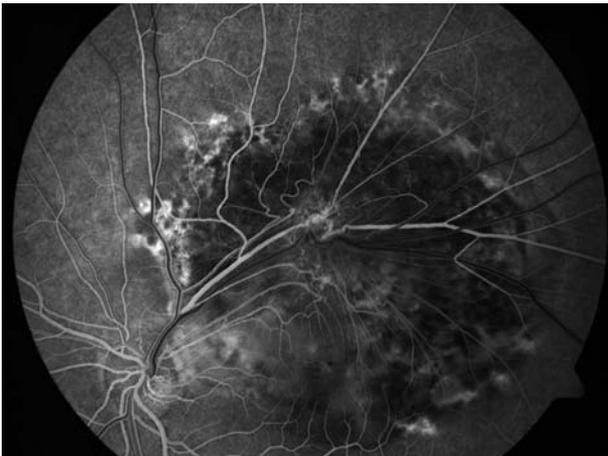
This is an interesting case with what I believe is a unique constellation of findings in a relatively young individual: a combined hamartoma of the retina and RPE associated with a staphyloma. The question in my mind is whether the chronic hamartomatous changes resulted in the staphyloma. Also, is the tractional macular detachment unique in that there is an associated staphyloma?

In other words, if the patient did not have the staphyloma, would we not see the macular detachment as observed on OCT? The answer to this question is probably not, just epiretinal proliferation, retinal thickening, and macular edema. Shields et al<sup>1</sup> have nicely characterized some of the OCT observations seen with combined hamartomas, but did not observe any patients with tractional retinal detachments. Review of the superior OCT scan shows an epiretinal membrane, thickening and disruption of the retina, and cystic changes within the retina. The inferior retina is somewhat normal, although detached from the RPE. As far as management is concerned, there have been mixed results with removing epiretinal membranes in these patients. With preoperative OCT, better surgical

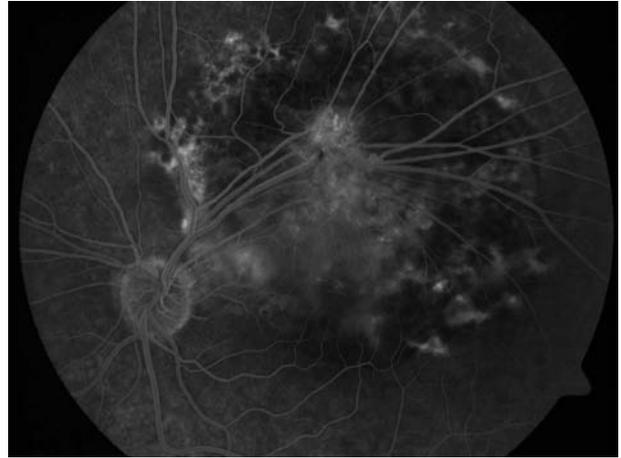


**Fig. 1.** Color photograph of the left eye showing the circular tractional retinal detachment with underlying RPE hyperpigmentation. The center of the lesion shows the retinal vessels gathered up into the contracted macular pucker.

planning is now possible. During the past 5 years, there has been significant improvement in vitreoretinal instrumentation and techniques (associated with understanding of the roles of the posterior hyaloid, the variety of epiretinal membranes that can be observed, and the role of the internal limiting membrane in abnormalities of the vitreoretinal interface). Therefore, my bias, particularly in a case such as this where there is a documented decline in vision, is to attempt to remove the epiretinal membrane and restore the anatomy of the macula. Therefore, pars plana vitrectomy, membrane peeling, and in this case, instillation of an expansile gas to help flatten the macula would be the surgical procedure I would perform. Of course, I could be completely wrong and this could simply be an epiretinal membrane. The



**Fig. 2.** Fluorescein angiogram of the left eye shows normal filling of retinal vessels with blockage of the choroidal fluorescence and patchy hyperfluorescence at the edge of the lesion.

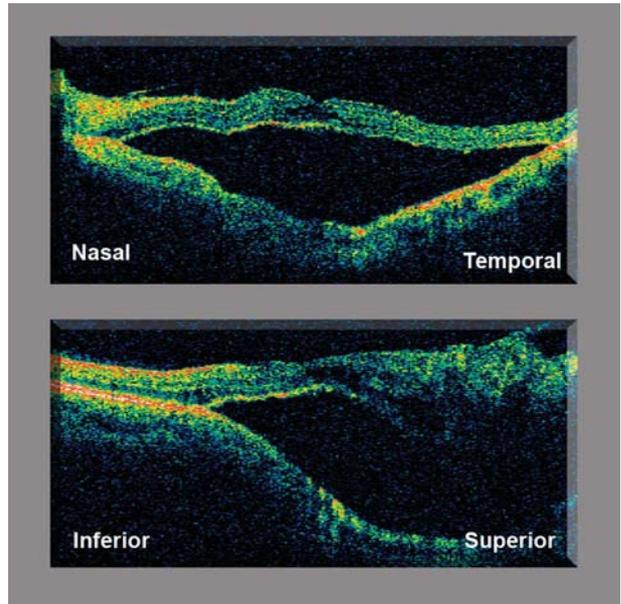


**Fig. 3.** Fluorescein angiogram of the left eye at 2 minutes shows leakage from tortuous retinal vessels in the center of the lesion. The edges of the lesion show increasing hyperfluorescence, but the choroid remains dark in the center of the lesion.

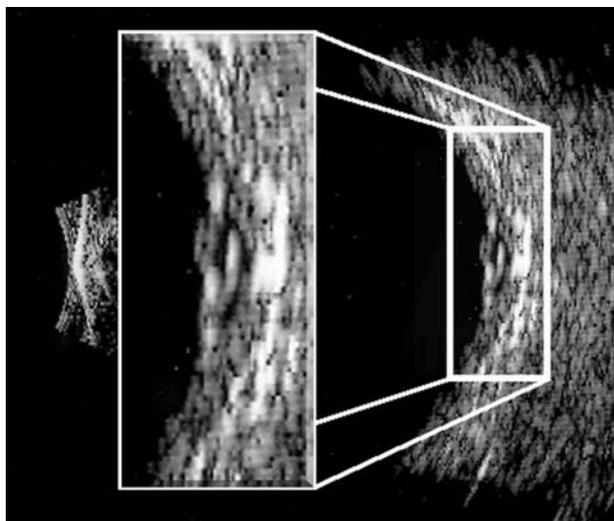
distinction between a combined hamartoma of the RPE and retina and an epiretinal membrane can be difficult, but the treatment would be the same!

**Dr. Jorge Guillermo Arroyo (Boston, Massachusetts):**

The author presents an intriguing case of a 42-year-old man with darkening central vision for 6 months, a 5-year history of distortion in vision, and a 10-year history of mild loss of vision in the left eye. Six weeks



**Fig. 4.** Horizontal (350°, 8 mm) and vertical (90°, 8 mm) line scan through the lesion shows subretinal fluid. The back of the eye curves back away from the retina and the retina is also elevated. The fovea is involved.



**Fig. 5.** A vertical axial B-scan through the lesion shows a small staphyloma under the retinal detachment.

before presentation, the patient's vision was measured at the 20/30 level with a mildly hyperopic refraction.

The fundus photograph of the left eye shows a 9-mm oval pigmented subretinal lesion with a mottled hypopigmented and well-defined edge associated with a focal area of whitish epiretinal membrane along the superotemporal arcade. Of note, there are retinal folds into the fovea from this focal area of epiretinal membrane with loss of choroidal detail in the inferior and nasal aspects of the macula suggesting macular detachment. A scalloped edge of the epiretinal membrane or posterior hyaloid is seen between the optic nerve and the center of the lesion.

The fluorescein angiogram demonstrates hypofluorescence in the central areas of this lesion, which remained dark in the later frames of the angiogram consistent with blockage by the hyperpigmented RPE. The edges of the lesion are hyperfluorescent early in the angiogram and stain late in the angiogram, consistent with staining. Finally, the vessels within the area of epiretinal membrane contraction leak late in the angiogram.

An ocular coherence tomogram shows a modest posterior staphyloma associated with subretinal fluid involving the fovea. One frame shows significant retinal thickening and distortion secondary to traction.

This case seems to be most consistent with trauma-induced focal vitreoretinal traction causing a lesion simulating a combined RPE and retinal hamartoma. There is evidence of long-standing but subclinical retinal traction, which may have resulted in RPE hyperplasia throughout most of the lesion. More recently, increased retinal traction has resulted in involvement of the macula and decreased vision.

Of note, there are two small areas of retinal whitening consistent with cotton wool spots within the area of retinal contraction. Cotton wool spots in association with epiretinal membranes have been described previously and typically resolve within 1 week after membrane peeling.

As opposed to a combined RPE and retinal hamartoma where surgery has a guarded prognosis, surgical peeling of the epiretinal membrane with relief of the vitreoretinal traction would be expected to reattach the macula and improve vision. At the time of surgery, the scalloped edge of the membrane or posterior hyaloid seen superior to the optic nerve may allow easier delineation of the plane between the retina and epiretinal membrane. Given the fact that the patient does have a small staphyloma in this area, avoidance of a retinal break during peeling is critical. Although most macular retinal breaks that are free of traction are relatively benign, macular breaks overlying staphylomas are a cause for concern. Thus, to prevent iatrogenic retinal breaks in this case, a bimanual technique using end-grabbing forceps and a lighted-retinal pick (or a retinal pick and chandelier-type illumination) would provide the most controlled method of peeling this membrane off the retina.

If no break is created during peeling of the epiretinal membrane, no intraocular tamponade would be needed. However, if a break developed, drainage of the subretinal fluid through the break, light laser treatment, and a fluid air exchange with face-down positioning would be advisable.

#### Editor's Note:

Dr. Steven Cohen has presented a 42-year-old man with a 5-year history of decreased and distorted vision in the left eye. Fundus examination showed a pigmented lesion with overlying traction detachment. Beneath this lesion was a staphyloma.

We asked Drs. Elias Reichel and Jorge Arroyo to consult on this case. Dr. Reichel feels this lesion is a combined retinal RPE hamartoma and notes that traction retinal detachment has not been previously described. He feels that given the documented decrease in vision, epiretinal membrane dissection should be attempted, despite the mixed visual results associated with vitrectomy for combined hamartomas.

Dr. Arroyo feels that this lesion is trauma-induced, resulting in a lesion simulating a combined hamartoma. He feels the longstanding but subclinical retinal traction detachment may have resulted in the RPE hyperplasia. He, too, is in favor of surgery, because removal of the epiretinal membrane would be expected to have a better chance if the lesion was, indeed, a combined hamartoma.

I have asked my associate, Dr. Brandon J. Lujan, to comment on the OCT presented in this case.

**Dr. Brandon J. Lujan (San Francisco, California):** \_\_\_\_\_

Although the Stratus OCT suggests the presence of posterior staphyloma, this cannot be definitively stated. Stratus is based on time-domain OCT technology, which uses a moving reference arm to assess reflectivity at consecutive depths. The stratus software then needs to reconstruct and align each adjacent A-scan. For this reason, pigment epithelial detachments on stratus often look “inverted.” The absolute position of the RPE and true geometry of the posterior pole could be determined, however, with spectral domain OCT systems, which move much faster given their ability to obtain an entire A-scan at once. The traction bond present superiorly may be solely responsible for this configuration, but the possibility of a staphyloma cannot be excluded.

On a personal note, I have seen a number of lesions that look similar to the one presented. I believed them to be combined retinal-RPE hamartomas. But, occasionally, some of these lesions have a paucity of retinal vascular tortuosity and/or glial membrane formation. Such lesions have been described as having unilateral, idiopathic leopard-spot lesions of the RPE.<sup>2</sup> Recently

this entity has been rebranded unilateral RPE dysgenesis.<sup>3</sup> Indeed, an OCT included in the updated article concerning this entity somewhat resembles the one presented in this case.

We thank Dr. Cohen for presenting this case, and Drs. Reichel, Arroyo, and Lujan for their comments.

### References

1. Shields CL, Mashayekhi A, Dai VV, Materin MA, Shields JA. Optical coherence tomographic findings of combined hamartoma of the retina and retinal pigment epithelium in 11 patients. *Arch Ophthalmol* 2005;123:1746–1750.
2. Cohen SY, Massin P, Quentel G. Unilateral, idiopathic leopard-spot lesion of the retinal pigment epithelium. *Arch Ophthalmol* 2002;120:512–516.
3. Cohen SY, Fung AE, Tadayoni R, et al. Unilateral retinal pigment epithelial dysgenesis. *Am J Ophthalmol* 2009;148:914–919.

---

**RETINA®**, *The Journal of Retinal and Vitreous Diseases*, encourages readers to submit **Diagnostic and Therapeutic Challenges** to retina@retinajournal.com. Cases for the Diagnostic and Therapeutic Challenges section should include a detailed history of the patient, the diagnosis, the workup, the management, and finally, the question or questions that the submitter wishes to have answered by the consultants.