A 52-Year-Old Male with a Peculiar Retinal Lesion

Wendy Shieh, MD; Thomas K. Krummenacher, MD

Introduction:

A 52-year old man with an ocular history of LASIK surgery OU presented for an evaluation of a retinal lesion in the left eye. Other than presbyopia-related symptoms, the patient denied any recent vision changes. His medical history is only significant for hypertension and asthma.

Examination:

The patient's best-corrected visual acuity(BCVA) was 20/25 in the right eye and 20/20 in the left eye. Pupillary exam did not reveal an afferent pupillary defect and intraocular pressures were within normal limits. Slit-lamp examination showed a normal anterior segment exam bilaterally. No abnormalities were noted in the posterior pole of the right eye. Dilated fundus exam of the left eye showed rare drusen in the macula and a solitary red-orange mass with associated feeder and draining vessels in the periphery (Figure 1). There was no evidence of any surrounding exudation or excessive subretinal fluid.

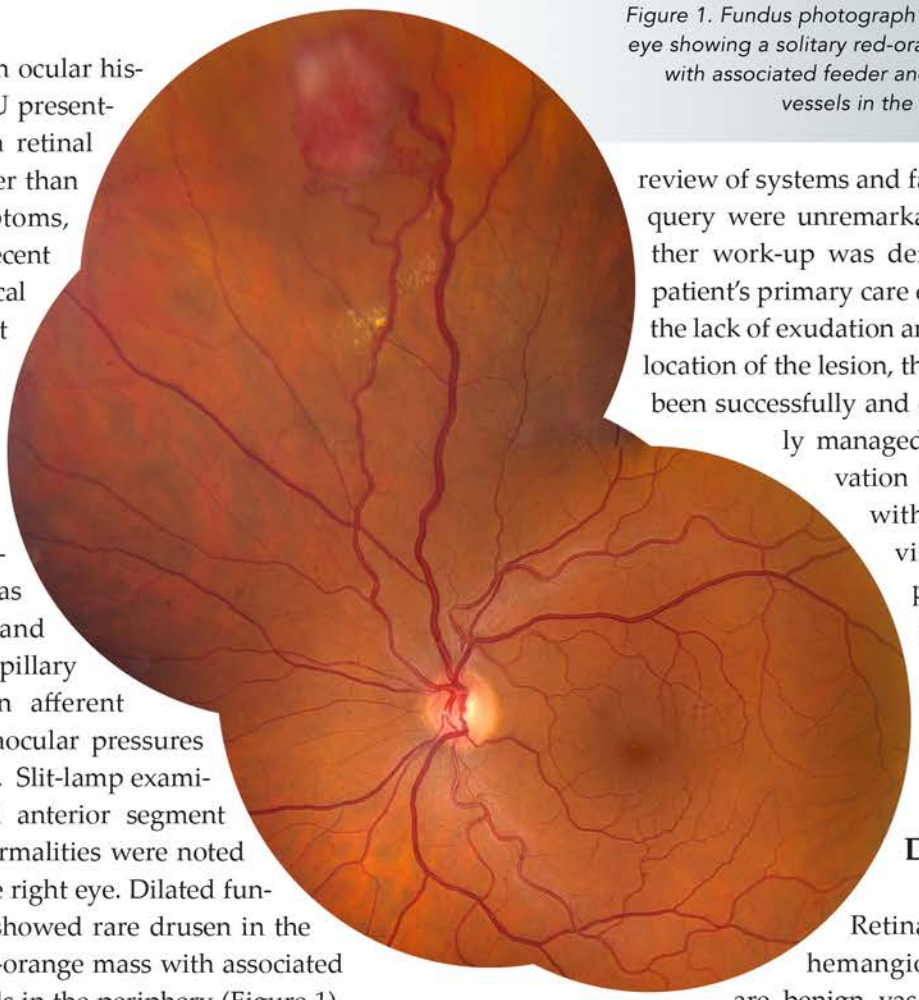


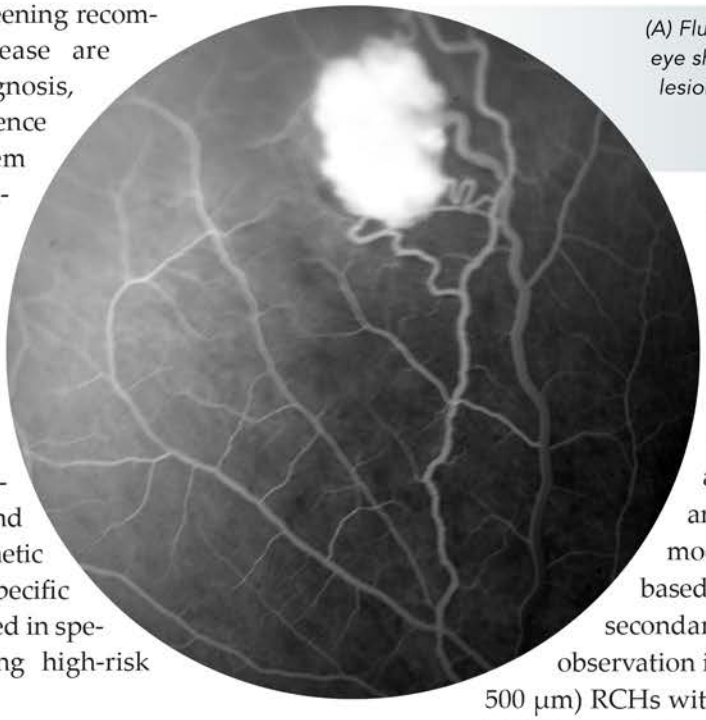
Figure 1. Fundus photograph of the left eye showing a solitary red-orange mass with associated feeder and draining vessels in the periphery.

Fluorescein angiography of the left eye demonstrated hyperfluorescence of the lesion with highlighting of the feeder artery as well as draining vein. There were no significant findings in the fellow eye (Figure 2). These clinical and angiographic findings were suggestive of a retinal capillary hemangioma and a more thorough history focused on characteristics of von Hippel-Lindau (VHL) disease was performed. Fortunately, a complete review of systems and family history query were unremarkable and further work-up was deferred to the patient's primary care doctor. Given the lack of exudation and peripheral location of the lesion, the patient has been successfully and conservatively managed with observation for 8-months without any visual complaints or complications.

Discussion:

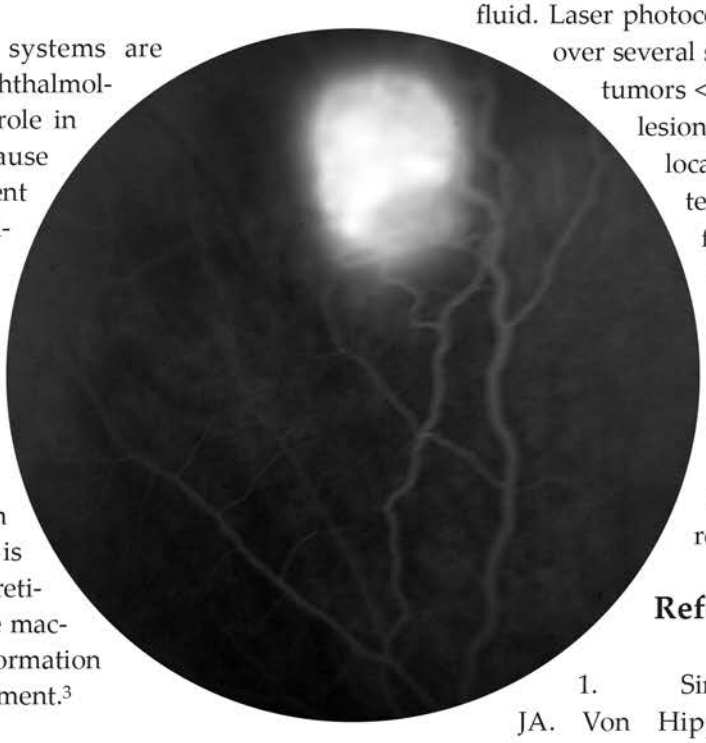
Retinal capillary hemangiomas (RCH) are benign vascular tumors of the retina and can occur either sporadically or as an ocular manifestation of VHL disease.[1] VHL is an autosomal dominant disease that has high age-dependent penetrance. The affected gene in VHL disease is a tumor suppressor gene on chromosome 3, where mutation leads to abnormal degradation of hypoxia inducible factor (HIF). The presence and activation of HIF stimulates proangiogenesis factors including vascular endothelial growth factor (VEGF), platelet-derived growth factor, erythropoietin, transforming growth factor and also affects extracellular matrix remodeling and

resistance to apoptosis.[2] Screening recommendations for VHL disease are based on the clinical diagnosis, which is defined as the presence of central nervous system (CNS) or retinal hemangioblastomas, a single CNS or retinal hemangioblastoma with an associated with visceral manifestation (pancreatic, renal or hepatic cysts, pheochromocytoma, or renal cancer), or any of the previously mentioned pathologic findings and a positive family history. Genetic testing in VHL is highly specific and sensitive and is conducted in specific circumstances involving high-risk individuals.[3]



(A) Fluorescein angiography of the left eye showing hyperfluorescence of the lesion with highlighting of the feeder artery as well as draining vein. (B) Late image of the lesion.

Although multiple organ systems are affected by VHL disease, ophthalmologists often play a pivotal role in diagnosing patients because RCHs are the most frequent and among the earliest manifestations of the condition.[1] Classically described as an orange-red retinal mass that can vary in size with dilated feeder vessels, RCHs can be located in the peripapillary region or in the retinal periphery. Vision loss associated with RCH is due to development of subretinal fluid, exudate within the macula, epiretinal membrane formation or a tractional retinal detachment.[3]



Solitary RCHs may also occur sporadically but are clinically indistinguishable from those found in patients affected by VHL. In a study by Singh et al., the authors report that the median age of RCH diagnosis in patients with VHL disease was approximately 19 years earlier than those without VHL disease. Of the 50 patients who initially presented with a solitary RCH, only 4 patients developed new tumors over a median follow-up of 6.4 years and were subsequently identified to have VHL disease.[4]

Treatment:

Various approaches have been described in the management of RCHs and include observation, laser photocoagulation, cryotherapy, plaque radiotherapy, and surgery. The treatment modality is often determined based on the size, location, and secondary effects of the RCH. Careful observation is appropriate in small (up to 500 μm) RCHs without exudation or subretinal fluid. Laser photocoagulation, which is applied over several sessions, is most effective for tumors < 1.5 mm but may be used in lesions up to 4.5mm. For anteriorly located RCHs (>3.0mm diameter) with associated subretinal fluid, treatment with cryotherapy is preferred over photocoagulation.[5] In advanced cases, particularly patients with severe exudative or tractional retinal detachments, pars plana vitrectomy may be required.[6]

References:

1. Singh AD, Shields CL, Shields JA. Von Hippel-Lindau disease. Surv Ophthalmol. 2001 Sep-Oct;46(2):117-42.

2. Haddad NM, Cavallerano JD, Silva PS. Von hippel-lindau disease: a genetic and clinical review. Semin Ophthalmol. 2013 Sep-Nov;28 (5-6):377-86.

3. Webster AR, Maher ER, Moore AT. Clinical characteristics of ocular angiomatosis in von Hippel–Lindau disease and correlation with germline mutation. Arch Ophthalmol. 1999 Mar;117(3):371-8.

References: (cont.)

4. Singh AD, Nouri M, Shields CL, et al. Retinal capillary hemangioma: a comparison of sporadic cases and cases associated with von Hippel-Lindau disease. Ophthalmology 2001;108(10):1907-11.

5. Singh AD, Nouri M, Shields CL, et al. Treatment of retinal capillary hemangioma. Ophthalmology. 2002 Oct;109(10):1799-806.

6. Gaudric A, Krivosic V, Duguid G, et al. Vitreoretinal surgery for severe retinal capillary hemangiomas in von Hippel-Lindau disease. Ophthalmology. 2011 Jan;118(1):142-9.

UPCOMING EVENTS FOR 2018

For more information about any or all of these events. . .

Contact Jen at Jennifer.Freeman@rc-stl.com

March 10, 2018

12th Annual RRDF Trivia Night

April 28, 2018

Spring Retina Update

September 8, 2018

Midwest Ophthalmologic Symposium

Case of the Month Supported by:

RRDF & Retina Research & Development Foundation

Want to see past issues of the Case of the Month?
Visit our website at tri-stl.com

THE RETINA INSTITUTE

1600 S. Brentwood Blvd., Suite 800
St. Louis, MO 63144
(314) 367-1181
tri-stl.com

Kevin J. Blinder, MD
Sabin Dang, MD
Nicholas E. Engelbrecht, MD
M. Gilbert Grand, MD
Daniel P. Joseph, MD, PhD
Thomas K. Krummenacher, MD
Richard J. Rothman, MD
Gaurav K. Shah, MD
Bradley T. Smith, MD
Matthew A. Thomas, MD