History and Chief Complaint

A 15-year-old white male presented with symptoms of floaters, intermittent headaches while reading, and mildly blurred vision for 2 months. This was the first ocular exam not given by his pediatrician. Both family and past medical histories were unremarkable with the exception of an uncle with glaucoma.

Clinical Findings

- VA (SC): 20/60 R, 20/20 L (With lenses best corrected to 20/30 R)
- Goldmann IOP: 15 R, 15 L

The external exam, including pupils and confrontation visual fields, was within normal limits.

Optomap

The composite right fundus image reveals three peripheral orange-red round lesions: temporal (2.5 DD in size), nasal (1 DD), and superior nasal (3 DD). All three, even the smallest one in the nasal fundus, appear to have a large artery entering the lesion and a very large tortuous vein draining it. In the macula, white deposits form an incomplete radial pattern. The vessels are most obvious in the green separation view, whereas the lesions appear to glow in the red separation view. The left fundus image reveals a single nodular protrusion from the inferior nasal disc about one-fifth of a disc diameter in size. This lesion is visible in both the green and red separation views. No other abnormalities were visualized.

Differential Diagnosis

The large orange-red lesions are blood vessel tumors. The dilated and tortuous vessels entering and exiting the tumors are feeder vessels and, in combination with the lesions, are rather typical of retinal capillary hemangiomas in von Hippel Lindau (VHL) syndrome. Note that arterial venous malformation (AVM) has similar appearing vessels but no tumor. The incomplete macular radial pattern is due to hard exudates in Henle's fiber layer. The macula star (in this case not complete) is due to leakage from the lesions. The macula often acts as a “sink” and lipid accumulation in the macula occurs in multiple retinal vascular disorders. The core of the hemangioma is composed of tortuous, large diameter capillaries lined by normal endothelium.

Disposition and Follow-Up

A full body work-up for similar lesions in the brain, spinal cord, and abdominal cavity is recommended. The presence of similar lesions elsewhere confirms the diagnosis of VHL, as it did in this case. A retinal specialist should be consulted, a fluorescein angiogram obtained, and treatment for the lesions should be considered. Historically, laser and cryotherapy have been utilized but today other modalities should at least be considered. These include PDT and anti-VEGF (vascular endothelial growth factor) intravitreal injections. Since regrowth of tumors is common, therapy may need to be repeated. Our patient underwent successful photocoagulation and cryotherapy on all lesions in the right eye with some improvement in vision after 4 weeks. Observation was elected for the optic nerve lesion in the left eye at this time. This patient will undergo lifelong MR imaging to ensure early detection of recurrent intracranial and systemic lesions. The neurosurgeon elected for early treatment of the cerebellar hemangioma that was discovered during the full body work-up and prognosis is good for a complete recovery. VHL is typically autosomal dominant in inheritance pattern but in this case no other family member was found to have any lesions at present. Without a family history such as in this case, the incidence of VHL drops from one in 50,000 to about 1 in nearly 5 million. As Dr. Lou Catania has often said, “it ain’t rare if it’s in your chair”—and it was in Dr. Walborn’s chair!

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