Case Report Submission
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Racemose Angiomatosis: A Rare Anomaly

Abstract
A 56 year-old male’s complaint of decreasing vision more pronounced in one eye leads to the unearthing of a rare, congenital ocular arterio-venous malformation of potentially life-threatening proportions.

I. Case History
- Patient demographics: 56 year-old white male
- Chief complaint: A 56 year-old gentleman reported that he had not been able to see clearly at distance (for a year) or up close (for a few years) with either eye. He also stated that he could not see well in the dark (for a year) and that he had issues with glare when driving at night. Over-the-counter reading glasses (+2.75) improved his vision both during the day and at night.
- Ocular, medical history:
  - Ocular: The patient’s last eye exam was ten years ago, during which he was given +1.25 reading glasses and informed that he had “something” in the back of his left eye. No further testing was done because of his lack of health insurance. He noticed that his vision in the left eye had gradually decreased since then, especially over the last year. Before that eye appointment, he had not noticed his decreased vision in the left eye. The patient reported that he had had no ocular injuries, surgeries, family history of eye diseases, or anything termed a “lazy eye”.
  - Medical: No significant findings.
- Medications: None reported.
- Other salient information: None reported.

II. Pertinent findings
- Clinical:
  The patient’s best-corrected acuities were 20/20 OD through a refraction of +3.00-0.50x105 and 20/30- OS through a refraction of +3.50-0.50x105, with an add of +2.25 OU. His pupils were equally round and reactive to light, and there was no afferent pupillary defect observed. Cover test revealed ortho alignment at distance and near. Confrontation fields were full to finger counting in each eye, and extraocular muscle movements were smooth and full in all positions of gaze OU. Intraocular pressures measured with Goldmann applanation tonometry at 11:20AM were 20mmHg OD and 20mmHg OS. Slit lamp examination was significant for pingueculae OD and OS, arcus OD and OS, grade 4 angles OD and OS (by Van Herick measurement), and grade 1+ nuclear sclerotic cataracts with mild posterior subcapsular cataracts OD and OS. Dilated examination revealed optic nerve heads with cup-to-disc ratios of 0.45h/0.4v OD and OS. The optic nerve head in both eyes appeared pink with distinct margins. Vasculature in the right eye had an arterio-venous ratio of 2/3 with no apparent pathology. Vasculature evaluation of the left eye revealed tortuous and dilated vessels emanating from the optic nerve head, encased in white fibrotic lesions, extending in a
corkscrew-fashion from the optic nerve head temporally and superiorly beyond the macula. The macula in the right eye was clear and flat, and the macula in the left eye was obscured by a large, white lesion and a linear pre-retinal hemorrhage slightly temporal to the fovea. A few areas of hyperpigmentation (vs. subretinal hemorrhaging) were located centrally in the macular lesion. The whitish areas appeared more yellowish in color as they progressed temporally. The vitreous in the right eye was clear, and there was a posterior vitreous detachment in the left eye. The peripheral retina in each eye appeared flat and healthy, with no breaks or detachments.

-Physical: No significant findings.
-Laboratory studies: Basic lab studies to clear this patient for magnetic resonance imaging was performed with normal results.
-Radiology studies:
  1) Optical coherence tomography of the macula of the right eye revealed normal findings. OCT of the macula of the left eye revealed thickening and uneven elevation of the retina surrounding the fovea. The RPE layer also had an irregular appearance with missing areas; however the area under the fovea was intact.
  2) The patient was referred for a high resolution OCT scan and A and B ultrasound scans at another facility to rule-out orbital arterio-venous malformation and any axial length discrepancy, respectively. (These results are not available at this time to the author due to access restrictions, but permission to release the records is in the process of being requested.)
  3) Fluorescein angiography testing revealed numerous abnormal arterio-venous shunts and late macular hyperfluorescence without choroidal neovascularization.
  4) MRI studies with and without contrast ordered to rule-out arterio-venous malformations in the central nervous system revealed normal anterior and posterior cerebral circulation without evidence of arterio-venous malformation. There was no evidence of thrombosis in the straight, sagittal, sigmoid, and transverse sinuses, through which there was normal flow.
-Others: Color fundus photos revealed the nature and extent of lesions OS.

III. Differential diagnosis
-Primary/leading: Racemose hemangiomatosis OS with evidence of previous retinal vein occlusion.
-Others: Von Hippel-Lindau syndrome (retinocerebellar capillary hemangiomatosis), retinal macrovessel, congenital retinal vascular tortuosity, Coats disease, retinal cavernous hemangioma, retinal hemangioblastoma, familial exudative vitreoretinopathy, retinal vein collaterals, circumscribed choroidal hemangioma, diffuse choroidal hemangioma (1,4)

IV. Diagnosis and discussion
-Elaborate on the condition:
  Racemose hemangiomatosis, also referred to as Wyburn-Mason syndrome, Bonnet–Dechaume–Blanc syndrome, and congenital unilateral retinocerebellar vascular malformation syndrome (CRC syndrome), is characterized by unilateral retinal vascular malformations with abnormal arterio-venous connections (1,2). The malformations can be found to differing degrees in various forms of the disease, from barely observable lesions to extensive lesions involving structures beyond the eye (1,2,3).

  It is a rare and congenital condition, with only approximately 120 documented cases in the literature (1,2). There is no documented pattern of inheritance (1,2).
Racemose hemangiomatosis can be diagnosed at any age, but is most commonly diagnosed in patients who are around twenty years old (1).

This condition can be generally stable and without consequence when confined to the retina; however, complications can include hemorrhaging, exudates, vessel occlusions, and neovascular glaucoma, at times resulting in severe vision loss (1). The anomalous vessels can regress or progress, and can extend to the orbit, optic nerve, chiasm, optic tract, midbrain, cerebellum, and face (2). Turbulent blood flow in the vasculature can lead to vessel wall damage, thrombosis, vessel blockage, and sclerosis (2). When vessels in the cerebral vasculature are involved, life-threatening neurological consequences of this condition can result (1, 2).

-Expound on unique features:

Despite the extent and location of the lesion observed, the patient was correctable to 20/30- in the left eye, demonstrating the resilience of the visual system. The relatively late age of diagnosis in this patient likely occurred not only due to his lack of financial resources, but also because of the surprising lack of subjective visual disturbance from this substantial congenital anomaly during his lifetime.

V. Treatment, management

-Treatment and response to treatment

There is no effective treatment for this entity, though current therapeutic strategies include endovascular, surgical, and radiation procedures to prevent and/or lessen damage stemming from this condition (1,2). Patients with Wyburn-Mason syndrome should be examined with MRI and angiography to rule-out involvement of the cerebral vasculature (2). Careful long-term monitoring of patients with this condition is indicated (2).

-Bibliography


VI. Conclusion

-Clinical pearls, take away points if indicated

1) Proper education of patients with this condition (or any other condition), is paramount to patient compliance and effective patient care, especially when further diagnostic testing is necessary and the patient has minimal subjective visual complaints.

2) Patients’ subjective experience of their quality of vision may not be as strongly correlated to the apparent level of pathology present upon examination as the examiner may expect.

3) The importance that patients place on obtaining satisfactory vision with optical correction should be given high priority in patient management decisions. Minimizing this need may be detrimental to patient compliance with management of their non-refractive conditions. This patient was not eligible to receive glasses at no cost through the VA system. Instead, he was given a prescription for glasses which he never filled, likely due to financial reasons, and
was subsequently lost to follow-up. It is arguable that had he received prescription glasses at no cost, he might have been more willing to comply with the recommended follow-up schedule.