Wyburn–Mason syndrome presenting with bilateral retinal racemose hemangioma with unilateral serous retinal detachment

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Wyburn–Mason syndrome is associated with unilateral retinal racemose hemangioma. Rarely, it presents with bilateral and symmetrical grade of malformation. We describe a 37-year old male, who presented with Wyburn–Mason syndrome presenting with bilateral but asymmetrical retinal hemangioma. The eye with advanced grade of hemangioma was complicated with exudation, intraretinal fluid, neurosensory detachment, and reduced vision. He was treated with one intravitreal injection of bevacizumab, after which both the intraretinal fluid and neurosensory detachment resolved. His vision improved and was maintained till 1 year of follow-up.

Key words: Asymmetric, bevacizumab, racemose hemangioma, Wyburn-Mason syndrome

Wyburn–Mason syndrome is a rare, congenitally acquired, vascular malformation.[1,2] It is characterized by direct artery-to-vein communication in retina and ipsilateral central nervous system.[1-5] Archer *et al.* classified the retinal arteriovenous malformations (AVMs), also called retinal racemose hemangioma, into three grades according to their severity.[3] Grade I lesions are characterized by AV communications with abnormal capillary plexus, whereas grade II lesions lack a capillary bed. In grade III lesions, distinction between artery and veins becomes impossible.[4]

The most characteristic feature of this malformation is its unilaterality.[1,5] Only few cases of bilateral involvement have also been reported in literature. All these cases presented with a symmetrical grade of retinal hemangioma.[1,5,6] We describe clinical features and management of a patient with Wyburn–Mason syndrome who presented with bilateral but asymmetrical grade of retinal racemose hemangioma.

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Case Report

A 37-year-old male presented with complaint of decreased vision in right eye for 4 months. His systemic history was unremarkable. Best-corrected visual acuity (BCVA) in right eye was 20/60 and left eye was 20/20. Ocular examination revealed round and normal reacting pupils with no relative afferent pupillary defect and parallel visual axes. Anterior segment examination in both the eyes was unremarkable. Posterior segment examination showed dilated and tortuous vessels with AV communication in both the eyes with hard exudates in the macular region in the right eye [Fig. 1a and b]. While the AVMs in right eye were of grade II, AVMs present in left eye were of grade I. The periphery was normal in both the eyes. Fluorescein fundus angiography (FFA) showed normal arm-to-retina time and rapid transit time in both the eyes, non-leaky retinal AV communications in the left eye [Fig. 1c], and late leakage in the perifoveal area in the right eye [Fig. 1d]. Magnetic resonance neurogram finding included tortuous vertebral arteries in close approximation with left-side complex of cranial nerves VII and VIII [Fig. 2a] and bilateral multiple tiny cavernous and capillary hemangiomas in supra- and infratentorial compartments with predominant involvement of brainstem and cerebellum [Fig. 2b]. Hence,

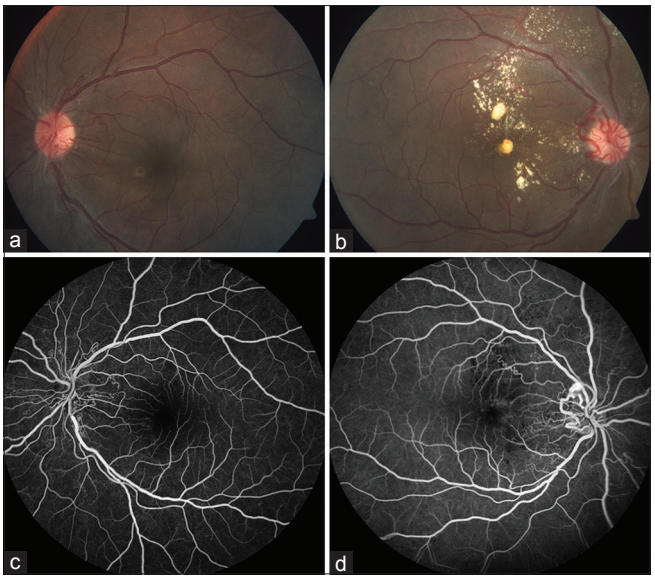

Figure 1: (a) Fundus photograph of left eye showing grade I AVM, (b) fundus photograph of right eye showing grade II arterioretinal venous malformation (AVM) and hard exudates, (c) FFA of left eye showing dilated non-leaky vessels, and (d) late phase of FFA of right eye showing giant vascular loops, direct arteriovenous connections, and late leakage in the perifoveal area

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the patient was diagnosed with incomplete Wyburn–Mason syndrome.[1]

Optical coherence tomography examination showed hyporeflective lesions in the inner retinal layers of the perifoveal and peripapillary region suggestive of dilated vessels in both the eyes [Fig. 3a]; hyperreflective lesions suggestive of hard exudates, intraretinal edema, and neurosensory detachment involving the fovea in the right eye [Fig. 3b]; while the left eye showed no macular edema [Fig. 3c].

The patient underwent one intravitreal injection of bevacizumab (IVB, 1.25 mg/0.05 mL) in his right eye. One month after the injection, BCVA in his right eye improved to 20/30. Both the intra- and subretinal fluid resolved and the exudates reduced. At 1-year follow-up, the patient maintained his BCVA at 20/30, while there was no recurrence of macular edema [Fig. 3d]. The vision did not improve completely because of the presence of hard exudate clumps in the outer layers of the fovea.

Discussion

We presented clinical findings of a patient with Wyburn–Mason syndrome and bilateral retinal racemose hemangioma. After a thorough search, we could find only eight cases of bilateral retinal hemangioma.[1,5,6] All the described cases had a symmetrical grade of retinal AVM in each eye. However, our patient presented with different grade of AVM in both the eyes. Second, these AVMs are usually stable without any signs of leakage on FFA.[1,2,4,5,7,8] However, in this case the eye with the higher grade of AVM was complicated by exudation, macular edema, and decreased vision, and the FFA showed late leakage in the perifoveal area.

The cause of macular edema in such cases in debatable. Soliman *et al.* suggested that the probable site of leakage may be either the capillaries adjacent to the AVM or the anastomosing vessels.[5] The venous pressure in the shunt vessels is exposed to high arterial pressure due to the absence of a normal capillary network.[1] This elevated venous transmural pressure causes excessive backpressure in the venous capillaries around the anastomosis. The raised capillary pressure decompensates the delicate balance maintained by the starling forces and the capillaries start leaking.[5,7]

Various modalities of treatment for the intraretinal and subretinal fluid in racemose hemangioma have been described by different authors [Table 1]. We investigated the role of intravitreal bevacizumab in the management of macular edema associated with retinal racemose hemangioma. Both intraretinal and subretinal fluid in our patient responded well to a single dose of IVB. The effect was maintained for 1 year, that is, the last date of follow-up. The exact mechanism through which bevacizumab reduces macular edema in these AVMs is unknown. It can be either due to the drug's ability to decrease the vascular permeability or due to its ability to reduce the number of dysplastic vessels.[2,9] The limitation of this case study is the absence of a long-term follow-up, which can help us understand the natural course of the patient. It would be interesting to know whether the AVMs in the second eye also become decompensated.

Conclusion

IVB seems to be a safe and effective option for the treatment of maculopathy associated with retinal racemose hemangioma.

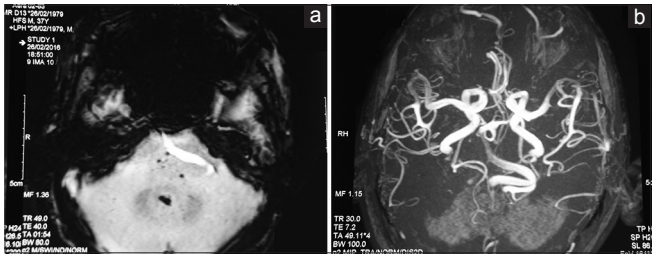

Figure 2: Magnetic resonance neurogram showing (a) tortuous vertebral arteries in close approximation with left-side complex of cranial nerves VII and VIII and (b) bilateral multiple tiny cavernous and capillary hemangiomas in supra- and infratentorial compartments with predominant involvement of brainstem and cerebellum

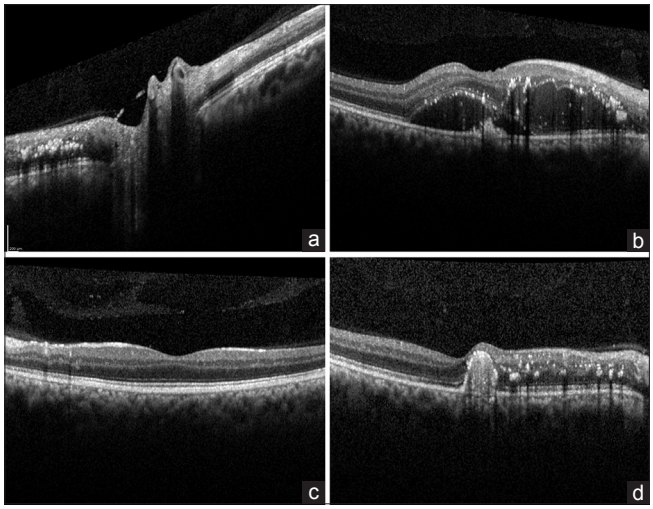

Figure 3: Ocular coherence tomography images of (a) left eye showing hyporeflective lesions suggestive of AV malformations in the inner retinal layers, (b) right eye at presentation showing intraretinal fluid and neurosensory detachment, (c) left eye with no macular edema, and (d) right eye 1 year after the injection

Table 1: Case reports describing the various modalities of treatment for maculopathy in racemose hemangioma

Author	Age	Presentation			Treatment	Result	
		Grade of AVM	BCVA	Type of fluid		Follow-up	BCVA
Chuang *et al.*[2]	37/F	Grade I	20/200	Intraretinal	3 IVB	2 years	20/20
Barreira *et al.*[4]	31/F	NA	20/40	Intraretinal	3 IVB	1 year	20/20
Soliman *et al.*[5]	57/F	NA	20/200	NA	2 Laser sessions	4 years	20/20
Onder *et al.*[7]	14/M	NA	20/30	Intraretinal, subretinal	1 PST TA (20 mg)	1 month	20/20
Winter *et al.*[8]	28/F	Grade I	20/60	Intraretinal	1 IVB	1 month	20/20

AVM: Arterio-venous malformation; BCVA: Best-corrected visual acuity; IVB: Intravitreal bevacizumab; PST: Posterior subtenon; TA: Triamcinolone acetate

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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