



Case report

Spectrum of peripheral retinal ischemia in Wyburn-Mason syndrome

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ABSTRACT

Purpose: We report two cases of Wyburn-Mason syndrome that illustrate the spectrum of peripheral retinal ischemia seen in this condition.

Observations: A 12-year-old female presented with a retinal arteriovenous malformation and sclerotic vessels associated with retinal ischemia on fluorescein angiography, as well as an ipsilateral ophthalmic arteriovenous malformation on magnetic resonance imaging. An 11-year-old male presented with retinal vascular engorgement and tortuosity along with a central retinal vein occlusion and secondary neovascularization.

Conclusions and Importance: Retinal ischemia in Wyburn-Mason syndrome is heterogeneous and may be progressive, with secondary complications that result in neovascularization. Furthermore, it is necessary to recognize that this is a systemic condition that requires neurological evaluation.

1. Introduction

Wyburn-Mason syndrome (WMS), also known as Bonnet-Dechaume-Blanc syndrome, is a rare, congenital, sporadic, unilateral condition characterized by retinal arteriovenous malformations (AVMs).¹ Ipsilateral AVMs of the cerebrum and face may be associated, although extraretinal findings are not required for the diagnosis. The location and severity of these findings determine the patient's clinical presentation. Ocular complications include vitreous hemorrhage, retinal vein occlusion, macular edema, both serous and rhegmatogenous retinal detachments, and neovascular glaucoma.²⁻⁷ Herein, we present two cases of Wyburn-Mason syndrome with peripheral retinal ischemia.

2. Findings

2.1. Case 1

A 12-year-old female with a history of Wyburn-Mason syndrome presented for a retina evaluation. Family ocular history was non-contributory. At initial presentation, Snellen visual acuity is 20/15 in the right eye and 20/200 in the left eye. Intraocular pressures were 15 mmHg in the right eye and 13 mmHg in the left eye. A relative afferent pupillary defect was noted in the left eye, along with a left exotropia.

Anterior segment exam of both eyes was unremarkable. Dilated

fundus exam of the left eye demonstrated an arteriovenous malformation with grossly dilated and tortuous vessels lacking capillary interposition obscuring the optic nerve and extending inferiorly (Fig. 1A). A few sclerotic vessels were also noted in the temporal periphery. Fluorescein angiography revealed an area of temporal peripheral non-perfusion (Fig. 1B). Further imaging with brain magnetic resonance imaging and magnetic resonance angiogram revealed a left ophthalmic AVM at the left suprasellar space and posterior intraconal fat extending to the left orbital apex with an associated superior hypophyseal aneurysm (Fig. 1C and D). The patient was scheduled for conventional cerebral angiography with possible embolization of the cerebral AVM.

2.2. Case 2

An 11-year-old male presented for a retina examination two weeks after experiencing sudden onset, persistent decreased visual acuity in the right eye, which was diagnosed as a central retinal vein occlusion (CRVO) with macular edema and a vitreous hemorrhage by an outside ophthalmologist. Family ocular history was non-contributory. At presentation, Snellen visual acuity in the right eye was 3/200 and 20/20 in the left eye, intraocular pressures were 15 mmHg in both eyes, and there was a right relative afferent pupillary defect.

Anterior segment exam was unremarkable in both eyes. Dilated fundus exam of the right eye demonstrated vitreous hemorrhage, along with sclerotic vessels and tortuous, dilated arteries and veins lacking an

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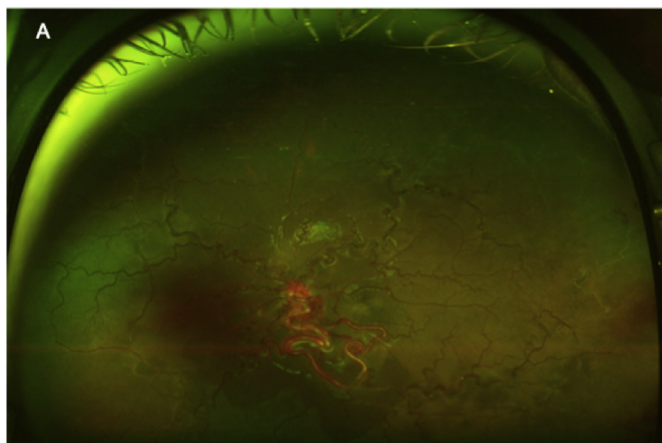


Fig. 1A. Wide field color fundus photograph of the left eye illustrating an arteriovenous malformation (AVM) with grossly dilated and tortuous vessels obscuring the optic nerve and extending inferiorly. Sclerotic vessels are also noted in the temporal periphery.

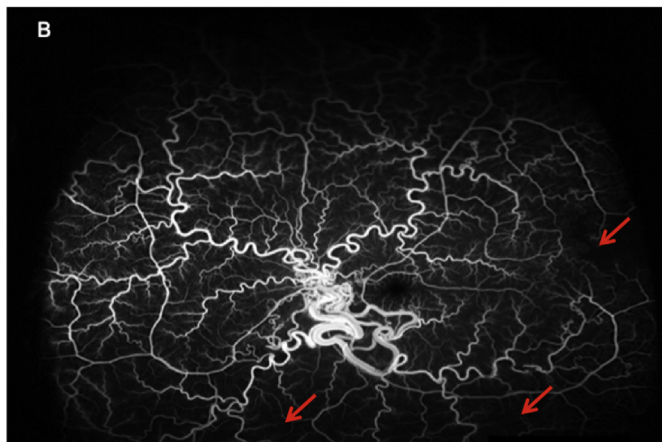


Fig. 1B. Fluorescein angiogram of the left eye demonstrating peripheral non-perfusion (arrows) in addition to the AVM.

interposing capillary bed (Fig. 2A). The left eye was normal. Optical coherence tomography (OCT) of the right eye revealed cystoid macular edema (Fig. 2B), and fluorescein angiography revealed peripheral non-perfusion circumferentially with multiple areas of capillary dropout and focal areas of late leakage along dilated vessels, along with peripheral leakage due to retinal neovascularization (Fig. 2C). Magnetic resonance imaging of the brain was negative for arteriovenous malformations or any other abnormalities. There were no cutaneous findings. Given that the patient was diagnosed with a CRVO, hematologic workup was pursued (cardiolipin antibodies, homocysteine, factor V, PTT, PT/INR, Protein C and S, antithrombin III antigen), which were all negative except for a marginally elevated phosphatidylserine IgM.

The patient was treated with a combination of photocoagulation in the region of avascular peripheral retina and intravitreal bevacizumab. 6 months after treatment with supplemental laser and 3 injections of bevacizumab, the cystoid macular edema improved, while the leakage along engorged vessels and peripheral neovascularization resolved (Fig. 2D and E).

3. Discussion

These two cases of Wyburn-Mason syndrome demonstrate the clinical spectrum seen in this condition, which may range from peripheral non-perfusion to severe ischemia resulting in retinal



Fig. 1C. Axial orbital magnetic resonance imaging scan and magnetic resonance angiogram demonstrating a left ophthalmic AVM at the left suprasellar space and posterior intraconal fat extending to the left orbital apex with an associated superior hypophyseal aneurysm (arrows).

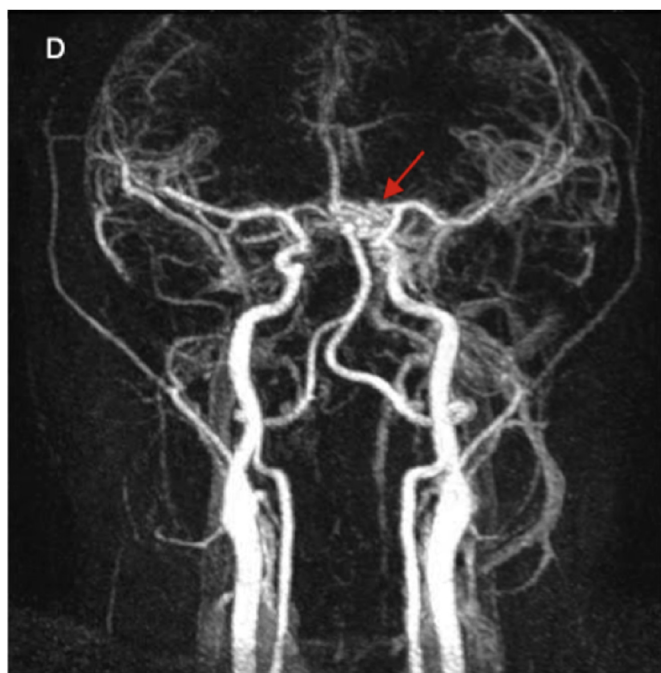


Fig. 1D. Axial orbital magnetic resonance imaging scan and magnetic resonance angiogram demonstrating a left ophthalmic AVM at the left suprasellar space and posterior intraconal fat extending to the left orbital apex with an associated superior hypophyseal aneurysm (arrows).

neovascularization. Classically, Wyburn-Mason syndrome has not been associated with retinal neovascularization, however a case of peripheral retinal neovascularization caused by Wyburn-Mason syndrome without angiographic evidence of a vein occlusion has been reported in the literature.⁸ In addition, Bloom et al.⁹ illustrated an AVM case complicated by retinal ischemia, sclerotic vessels, neovascular glaucoma, and

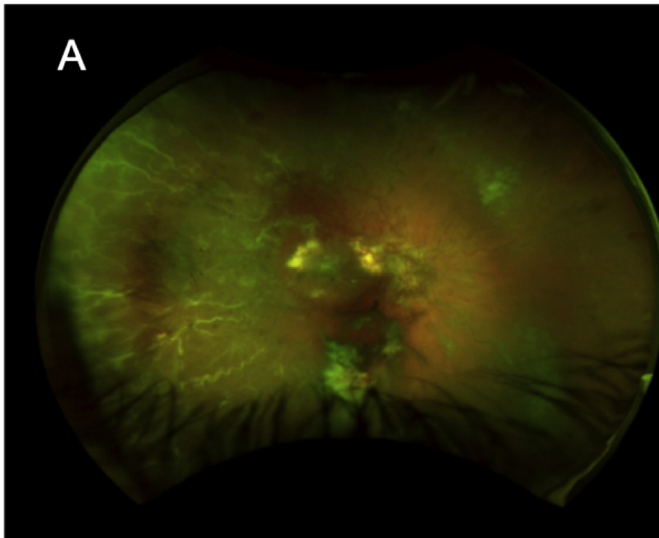


Fig. 2A. Wide-field color fundus photograph of the right eye demonstrating an AVM with engorged and tortuous vessels, a vitreous hemorrhage, and sclerotic vessels nasally.

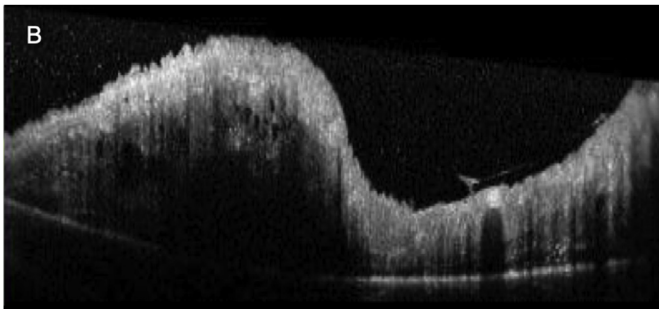


Fig. 2B. Optical coherence tomography of the right eye showing cystoid macular edema at presentation.

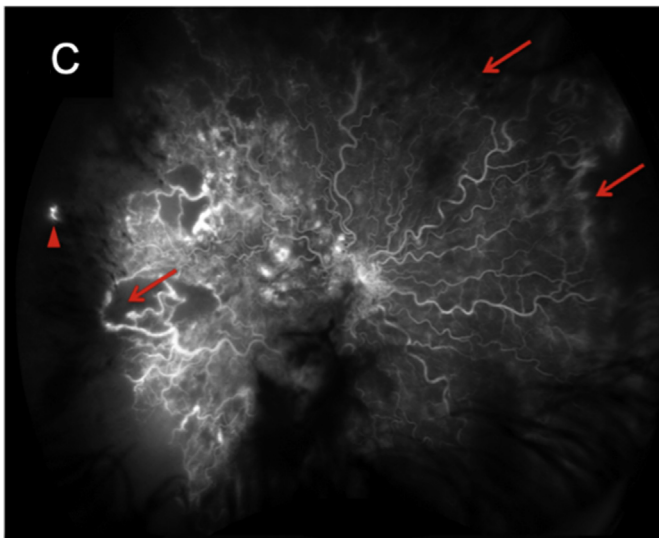


Fig. 2C. Fluorescein angiogram of the right eye revealing a blocking affect as a result of the vitreous hemorrhage, multiple areas of capillary dropout (arrows), and multiple hyperfluorescent areas along engorged vessels with focal leakage in the temporal periphery consistent with retinal neovascularization (arrow-head).

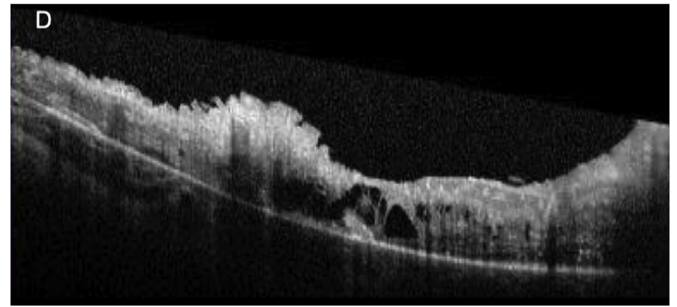


Fig. 2D. Optical coherence tomography of the right eye showing improvement of cystoid macular edema after 3 injections of bevacizumab and supplemental laser to the avascular retina.

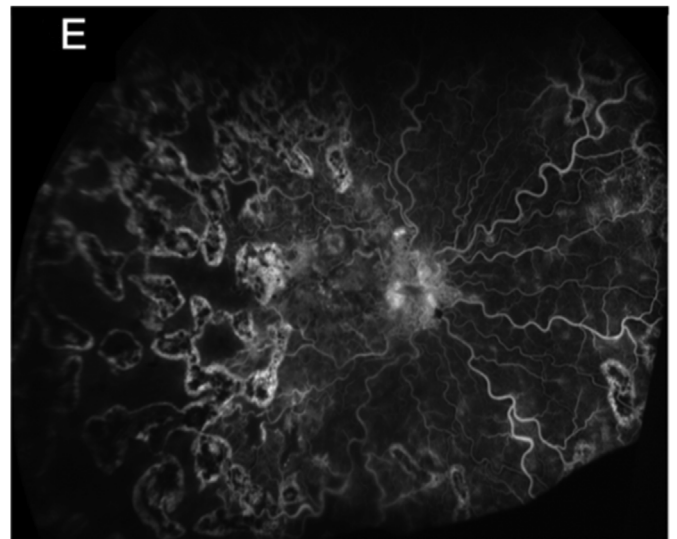


Fig. 2E. Fluorescein angiogram of the right eye revealing resolution of leakage along the peripheral engorged vessels and neovascularization 6 months after treatment with peripheral laser and intravitreal bevacizumab.

subsequent development of a tractional retinal detachment, suggesting retinal neovascularization as the underlying etiology. Several cases of neovascular glaucoma as a result of CRVO in the setting of retinal AVMs have been reported as well.⁵⁻⁷ Distinct mechanisms suggested as the underlying etiology of ischemia include direct compression of venous vasculature from the AVM, partial thromboses, or a “steal” phenomenon whereby blood is diverted from the lesion to other parts of the retina.⁸ Histopathologic studies on vascular changes in AVMs demonstrate a chronic fibrotic and thrombotic process of involved vessels as a result of turbulent blood flow due to a lack of capillary interposition.¹⁰ In agreement with previous theories on the pathophysiology of WMS, it is hypothesized that vascular compression and thrombosis of smaller vessels may eventually evolve into compression of the dilated central retinal vein at the level of the lamina cribrosa and subsequent ischemia, which may predispose to retinal neovascularization.¹

Case 1 is unique in that WMS without profound vaso-occlusive events may still have peripheral retinal ischemia. This is in comparison to case 2, in which vascular occlusion progressed to involve the central retinal vein, and subsequently resulted in ischemia severe enough to cause neovascularization. Hematologic workup was performed, as hypercoagulable conditions must be considered in the differential diagnosis of central retinal vein occlusion in a young patient with no known cardiovascular risk factors.¹¹ In addition, it is important to distinguish ischemic CRVO from non-ischemic CRVO prior to attributing retinal neovascularization to WMS, as ischemic CRVO may represent a potential etiology in and of itself. Ischemic CRVO is associated with retinal

neovascularization in 29% of cases within 12 months of vein occlusion, whereas the non-ischemic variety is not.¹² In this case, fluorescein angiography at one month after CRVO indicates an ischemic CRVO, which is the likely secondary etiology for the retinal neovascularization. Both cases may represent the progression of Wyburn-Mason syndrome from early capillary non-perfusion in case 1 to widespread ischemia resulting in retinal neovascularization after CRVO in case 2.

A thorough neurological assessment is important to evaluate for the systemic features of WMS. Magnetic resonance (MR) imaging of the brain and orbit may be considered as well as MR angiography or conventional cerebral angiography for additional information concerning the hemodynamics and arterial supply of intracranial AVMs.

4. Conclusions

Wyburn-Mason syndrome is a sporadic condition with a wide range of ophthalmic complications; it is important to consider neurological evaluation of patients with this condition. The two cases presented suggest that the retinal findings seen in Wyburn-Mason syndrome are progressive, and that close monitoring is important to detect worsening retinal ischemia, especially given the risk of neovascularization.

Patient consent

Consent to publish this case report was not obtained. This report does not contain any personal information that could lead to the identification of the patients.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

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