


# Peculiar aetiology for orbital apex syndrome: Wyburn-Mason syndrome as orbital apex lesion

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## ABSTRACT

**Background** Wyburn-Mason syndrome is a rare, non-hereditary congenital disease, belonging to the group of neurocutaneous syndromes with fewer than 100 cases reported since its first description in 1937.

**Case report** A young adult man was initially evaluated at the age of 2 years for proptosis and progressive visual impairment of the right eye, followed by impairment in ocular abduction, adduction and elevation as well as amaurosis. MRI revealed an expansive formation centred in the right orbit compromising conal spaces with distortion of eye muscles and optic nerve. The lesion extended through the superior orbital fissure into the right cavernous sinus and to the contralateral orbit. Despite embolisation, proptosis and oedema of the periorbital tissue continued to worsen. The combination of facial, ocular and intracranial vascular malformations and the exclusion of alternative aetiologies led to a diagnosis of cerebrofacial arteriovenous metameric syndrome (CAMS) 1 (Wyburn-Mason syndrome).

**Discussion** Important differential diagnoses are other CAMS, such as Sturge-Weber syndrome, as well as other conditions such as retinal cavernous haemangioma and vasoproliferative tumours. The optimal treatment regimen for severe cases of this syndrome is still unclear. Wyburn-Mason syndrome should be considered in patients presenting multiple arteriovenous malformations with orbital apex lesions.

## CASE REPORT

A young adult man was initially evaluated at the age of 2 years for proptosis and progressive visual impairment of the right eye. An erythematous supraorbital skin lesion was perceived at that moment. There was no family history of neurological disorders, and his parents were non-consanguineous. MRI revealed an arteriovenous malformation (AVM) involving the right cavernous sinus, which was treated with embolisation. Despite the procedure, proptosis and oedema of the periorbital tissue continued to worsen, progressing with severe visual loss. At the age of 12 years, he underwent a biopsy of the frontal, infraorbital and periorbital

subcutaneous tissue that was compatible with a cavernous angioma. The combination of facial, ocular and intracranial vascular malformations and the exclusion of alternative aetiologies led to a diagnosis of Wyburn-Mason syndrome. At the age of 18 years, he was evaluated at our institution. On neurological examination, there was marked proptosis with impairment in abduction, adduction and elevation as well as amaurosis of the right eye (online supplemental figure 1). Oedema of periorbital, frontal and zygomatic regions on the right was also seen. MRI revealed an expansive formation centred in the right orbit causing proptosis and compromising conal spaces with distortion of eye muscles and optic nerve. The lesion extended through the superior orbital fissure into the right cavernous sinus and to the contralateral orbit ([figure 1](#)). Intraocular bevacizumab was not considered for the right eye since it was a long-standing lesion, and he also had extrinsic compression of the optic nerve.

## DISCUSSION

Wyburn-Mason syndrome also known as Bonnet-Dechaume-Blanc syndrome or retino-encephalofacial angiomatosis is a rare, non-hereditary congenital disease, belonging to the group of neurocutaneous syndromes classified as cerebrofacial arteriovenous metameric syndromes (CAMSs), with fewer than 100 cases reported since its first description in 1937.<sup>1</sup> In this classification, there may be involvement of the hypothalamus and nose in CAMS1; of the occipital lobe, thalamus and maxilla in CAMS2; or of the cerebellum, pons and mandible in CAMS3 or even mixed phenotypic expressions, all with their own characteristics, natural history and symptoms.<sup>2,3</sup>

Wyburn-Mason syndrome usually affects the orbit, retina and brain.<sup>3</sup> Lesions may also

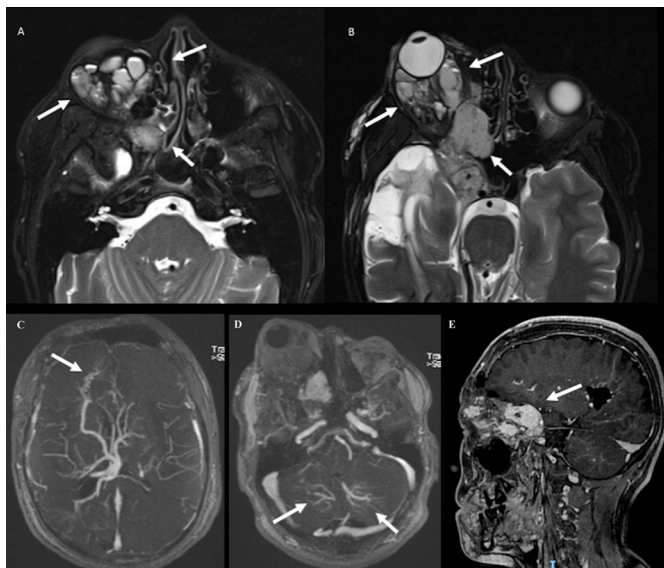


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**Figure 1** Brain MRI axial T2 scans (A, B) and postcontrast sagittal T1 (E), showing an expansive formation in the right orbit causing proptosis and extending to the cavernous sinus through the superior orbital fissure. Magnetic resonance angiography revealed developmental venous anomalies in the right frontal and temporal regions and cerebellar hemispheres (C, D).

affect the skin, maxilla, jaw, pharynx, oral and nasal cavities.<sup>4</sup> Aetiology is uncertain, but an embryonic defect is believed to result in premature dissemination of vascular cells from the cephalic mesoderm along the migratory route.<sup>3,4</sup>

Important differential diagnoses are other CAMS, such as Sturge-Weber syndrome, as well as other conditions such as retinal cavernous haemangioma and vasoproliferative tumours. The involvement of other facial and cranial structures typical of a metameric syndrome, in this case with the involvement of the orbits and their attachments and the brain, helps in the differentiation.<sup>2,3</sup>

Treatment of AVMs with endovascular techniques, surgery and radiosurgery is a possibility, but conservative treatment may be preferred due to the high risk of recurrence.<sup>5,6</sup> Bevacizumab, an anti-VEGF agent, has been shown to contain macular oedema and improve visual loss in a short-term follow-up.<sup>7</sup> There are no guidelines to treat severely affected patients like in the present case. More studies are needed to evaluate possible treatments for advanced Wyburn-Mason syndrome, including the possibility of using intravenous bevacizumab. Monitoring should be carried out to assess the appearance of lesions in other typical locations and contralateral involvement; however, more evidence is needed to define the ideal frequency and method.

The present study demonstrates that Wyburn-Mason syndrome should be considered as a differential diagnosis in cases presenting multiple AVMs with orbital apex lesions.

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#### REFERENCES

- 1 So J, Mishra C, Holman R. Wyburn-Mason syndrome - Statpearls - NCBI bookshelf. StatPearls; 2022. Available: <https://www.ncbi.nlm.nih.gov/books/NBK493218/> [Accessed 9 Jun 2023].
- 2 Jiarakongmun P, Alvarez A, Rodesch G, et al. Clinical course and angioarchitecture of cerebrofacial arteriovenous metameric syndromes. *Interv Neuroradiol* 2002;8:251-64.
- 3 Bhattacharya JJ, Luo CB, Suh DC, et al. Wyburn-mason or bonnet-dechaume-blanc as cerebrofacial arteriovenous metameric syndromes (CAMS): a new concept and a new classification. *Interv Neuroradiol* 2001;7:5-17.
- 4 Tomarchio S, Portale A, Praticò A, et al. Wyburn-mason syndrome. *J Pediatr Neurol* 2018;16:297-304.
- 5 Schmidt D, Pache M, Schumacher M. The congenital unilateral retinocephalic vascular malformation syndrome (bonnet-dechaume-blanc syndrome or wyburn-mason syndrome): review of the literature. *Surv Ophthalmol* 2008;53:227-49.
- 6 O'Loughlin L, Groves ML, Miller NR, et al. Cerebrofacial arteriovenous metameric syndrome (CAMS): a spectrum disorder of craniofacial vascular malformations. *Childs Nerv Syst* 2017;33:513-6.
- 7 Pangtey BPS, Kohli P, Ramasamy K. Wyburn-mason syndrome presenting with bilateral racemose Hemangioma with unilateral serous retinal detachment. *Indian J Ophthalmol* 2018;66:1869-71.