Introduction

Wyburn-Mason syndrome is a rare, congenital, non-hereditary entity characterized by orbital, intracranial, and facial arteriovenous malformations that are typically unilateral; atypical cases with bilateral manifestations are reported.1 Wyburn-Mason syndrome usually presents before the third decade of life2 with no sex or race predilection.3 In 1937, Bonnet et al.4 first described the combination of ipsilateral retinal and intracranial arteriovenous malformations. In 1943, Wyburn-Mason reviewed 27 cases of retinal arteriovenous anastomoses and found that 81% of the patients had comitant cerebral arteriovenous malformations.1 This condition results from a defect during embryonic development of the vascular mesoderm related to the visual pathways from the mesencephalon to the retina. The time of the insult to the embryonic tissue determines which structures are affected. We present a case of Wyburn-Mason syndrome with fatal outcome and briefly discuss the management and related literature.

Case report

We present the case of a 47-year-old man with Wyburn-Mason syndrome, which was diagnosed by his previous physicians. Intra-orbital and multiple facial and intracranial vascular malformations, predominately in the left frontal and temporal region, were identified (Figures 1a,b,2a). Prior to his presentation, numerous angioembolisations of the external carotid artery were performed to control repetitive bleeding episodes. His first intracranial haemorrhage occurred at the age of 7, and he developed left sided hemiparesis at the age of 28. Furthermore, the patient was treated previously for epilepsy related to multiple intracranial haemorrhages. MRI of the neurocranium revealed multiple haemorrhage-related intracranial lesions (Figure 2b). The patient suffered from persisting, sometimes massive bleeding episodes, most commonly from the medial canthal area of the left orbit. Ocular examination revealed vision loss of the left eye. Visual acuity of the right eye was 2/100 and there was no impairment of motility. Again, at the age of 49, the patient was treated with multiple angioembolisations and radiation therapy of the left orbit to control the persistent bleeding. At the age of 50, recurrent bleeding episodes required treated via surgical intervention with left orbit exenteration; this resection was required because the enlarged left ophthalmic artery fed the bleeding vascular malformation (Figure 3). After surgery, bleeding episodes were significantly reduced. At the age of 52 the
patient presented with complaints of acute sinusitis. CT scan showed an empyema of the left maxillary sinus (Figures 4a,b). Endoscopic intervention had to be aborted due to massive endonasal bleeding from the vessels feeding the arteriovenous malformation; the patient was subsequently treated via a transorbital approach.

The postoperative course was uneventful, and bleeding episodes related to the vascular malformations remained rare. At the age of 54 the patient died of cardiovascular disease; an autopsy was not performed.

**Discussion**

Wyburn-Mason syndrome is a rare disease associated with multiple arteriovenous malformations involving the brain, orbit, and face. The aetiology and risk factors of this syndrome are unknown and there is no sex or race predilection. The currently reported case represents the common characteristics seen with Wyburn-Mason syndrome, consisting of neurologic, ocular, orbital, and dermatological symptoms. Typical neurologic symptoms are headache, seizures, visual field loss, mental changes, neurologic deficits, and epilepsy. Our patient also suffered from hemiparalysis and epilepsy, which is the most common complaint in patients with arteriovenous malformations of the brain. Ophthalmic manifestations may include decreased visual acuity or visual loss, proptosis, optic atrophy, and abnormal dilatation of conjunctival and iris vessels. All these characteristics were found in the current case. Relative to orbital and intracranial malformations, cutaneous lesions are rare in Wyburn-Mason syndrome. Typical dermatological findings are angiomas ipsilateral to the affected eye, as found in the presented patient.

The prognosis of Wyburn-Mason syndrome varies depending on the size, location, growth, and extent of the lesions; these characteristics can be determined by computed tomography, magnetic resonance imaging, and angiography. Morbidity and mortality are secondary to the
The bleeding risk of arteriovenous malformations. In particular, intracranial malformation bleeding is crucial because of the risk of cerebral or subarachnoid haemorrhage, neurological deficits, and death. The patient described here suffered from multiple intracranial haemorrhages, which led to neurological deficits including epilepsy and hemiparesis.

Due to the heterogeneous presentation, management of Wyburn-Mason syndrome is difficult, and appropriate treatment remains controversial. Nonoperative management with close observation for changes in lesion size is the current standard of care. Therapy is typically reserved for symptomatic patients. When intervention is contemplated the risks and benefits of treatment must be carefully weighed against that of observation alone. A variety of treatment strategies for Wyburn-Mason syndrome are discussed in the literature, including surgical extirpation, radiotherapy, and endovascular embolization. With these techniques it is sometimes possible to control the symptoms of Wyburn-Mason syndrome. In the presented case, after multiple angioembolisations and radiation therapy, surgical intervention proved to be successful and led to a persistent decrease in bleeding complications. In addition to the typical problem of recurrent bleeding episodes, patients with Wyburn-Mason syndrome can also develop atypical complications, such as maxillary sinus empyema. These atypical complications can further complicate the management of these patients. In the current case, the sinus empyema was treated via a transorbital approach after exenteration of the orbit two years previously. The standard endonasal endoscopic approach was impossible due to massive bleeding of the endonasal arteriovenous malformation.

Conclusion

This case demonstrates that several different treatment strategies are often necessary to control the symptoms and atypical complications of Wyburn-Mason syndrome. In order to make an educated decision regarding therapy, a thorough analysis of existing malformations with special regard to associated clinical symptoms is necessary to improve quality of life. In the present case, surgical therapy was ultimately successful in reducing bleeding episodes.

References


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