Wyburn-Mason syndrome

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Bonnet, Dechaume, and Blanc were first to recognize AVM of face, retina, and brain in the year 1937 and Wyburn-Mason described in detail about this disease in 1943. In recognition of these authors, this disease is referred to as Bonnet-Dechaume-Blanc syndrome or Wyburn-Mason syndrome.

Wyburn-Mason syndrome, also known as Bonnet-Dechaume-Blanc syndrome, is a rare vascular disorder that affects the arteries and veins in the brain and the eyes. It is characterized by the presence of arteriovenous malformations (AVMs), which are abnormal connections between arteries and veins.

The exact cause of Wyburn-Mason syndrome is not fully understood, but it is believed to be a result of an embryological developmental abnormality that occurs during the formation of the vascular system.

Symptoms of Wyburn-Mason syndrome can vary widely depending on the location and severity of the AVMs, but they typically involve abnormalities in vision and neurologic function. These may include visual disturbances such as blurred vision, loss of visual acuity, and defects in visual fields. Neurologic symptoms can include seizures, headaches, and cognitive impairment.

There is no cure for Wyburn-Mason syndrome, and treatment is typically aimed at managing the symptoms and preventing complications. This may involve a multidisciplinary approach, including interventions such as laser treatment, embolization, or surgery to address the AVMs, as well as medications to manage seizures or other neurologic symptoms.

Overall, the prognosis for individuals with Wyburn-Mason syndrome is highly variable and dependent on the location and extent of the AVMs, as well as the severity of associated symptoms. Regular monitoring and follow-up care are typically recommended to manage the condition and prevent further complications.

Neurocutaneous disorder: racemose angioma (Wyburn-Mason syndrome): midbrain and retinal arteriovenous malformations.

When encountering patients with markedly dilated and tortuous retinal vessels, Wyburn-Mason syndrome (WMS) or racemous angiomatosis (phacomatosis) is commonly thought of as the archetypal entity that can produce these findings.

Kisilevsky et al. described a patient with Cantu syndrome with phenotypical findings identical to those seen in patients with WMS and want to highlight this as another entity that can present with tortuous

and dilated retinal vessels [1].

As with other intracranial and intraocular vascular lesions (e.g., arteriovenous malformation in Wyburn-Mason syndrome, retinal hemangioblastoma in von Hippel Lindau disease, and choroidal hemangioma in Sturge-Weber syndrome), the presence of a vascular lesion in either location should prompt evaluation for additional pathology [2].

Case reports

A 62-year-old man with a 30-year history of WMS with right basal ganglia and orbital AVMs and right eye blindness presented for new-onset left-sided vision loss. A pituitary adenoma was identified compressing the optic chiasm and left optic nerve. Magnetic resonance imaging and digital subtraction angiography studies were obtained for surgical planning, and the patient underwent an endoscopic transnasal transsphenoidal resection, with significant postoperative vision improvement.

Lessons: Given the variable presentation and poor characterization of this rare syndrome, patients with WMS presenting with new symptoms must undergo evaluation for growth and hemorrhage of known AVMs, as well as new lesions. Further, in patients undergoing intracranial surgery, extensive preoperative imaging and planning are crucial for safe and successful procedures [3].

References

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