

CASE REPORT

Uncommon Presentation of Wyburn-Mason Syndrome in A Pediatric Patient: A Case Report

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ABSTRACT

We report an uncommon presentation of Wyburn-Mason syndrome in a pediatric patient. An 11-year-old girl presenting with no light perception in her right eye. Examination revealed an exotropia and a large and tortuous retina vessel in the posterior part of her right eye. However, other examinations, including dermatological and neurological examination, are within normal limits. Magnetic Resonance Angiography (MRA) showed vascular malformation at the basal cistern that extends to the retrobulbar area. Therefore, a thorough examination of the eye and brain, including MRA, is mandatory in every case, even in pediatric with normal neurological examinations.

Keywords: Arteriovenous Malformation, Racemose hemangiomas, Wyburn-Mason syndrome

Racemose hemangiomas or mostly known as Wyburn-Mason Syndrome is a rare, congenital, non-familial, vascular malformation characterized by ipsilateral arteriovenous malformations (AVMs) in the midbrain, retina, and occasionally facial skin.^[1-3] Most Wyburn-Mason syndrome did not affect visual acuity in the pediatric age group.^[4] This case report wants to highlight a possible serious vision loss in a child with Wyburn-Mason Syndrome.

CASE HISTORY

An 11-year-old girl presented in our tertiary eye care facility with a blurry right vision, which she first noticed it around

three months ago. There was no history of trauma, previous ocular diseases, or any significant family history. The initial examination revealed a seven degrees exotropia without a perception of light in her right eye. Meanwhile, the visual acuity of her left eye was 6/6. The anterior segment, ocular adnexa, and intraocular pressure on both sides were normal. Large, tortuous, and dilated retinal vessel was present in the posterior part of her right eye (Figure 1). Her left eye was unremarkable. She neither has any cutaneous lesion nor any symptoms associated with neurologic deficit. Magnetic resonance angiography (MRA) revealed vascular malformation located at the basal cistern, reaching the right third paraventricular, and extending

to the retrobulbar area of the right eye, thus confirming the diagnosis of Wyburn-Mason syndrome (Figure 2a and 2b). The interventional radiology department has planned to do a digital subtraction angiography and an embolization procedure. Unfortunately, she was lost to follow-up.

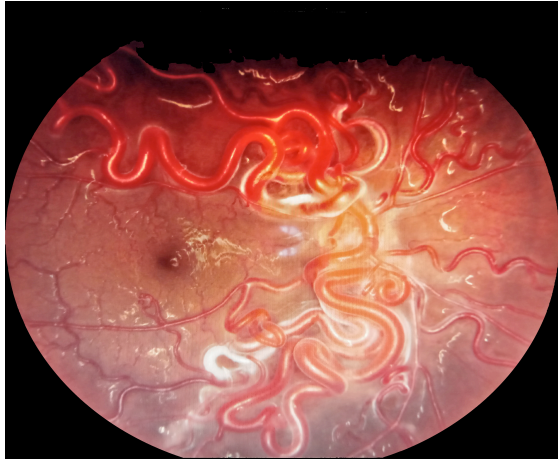


Figure 1. Fundus photograph of the right eye showed AVM with enlarged and tortuous vessels in all quadrants at the posterior pole of the retina without capillary networks.

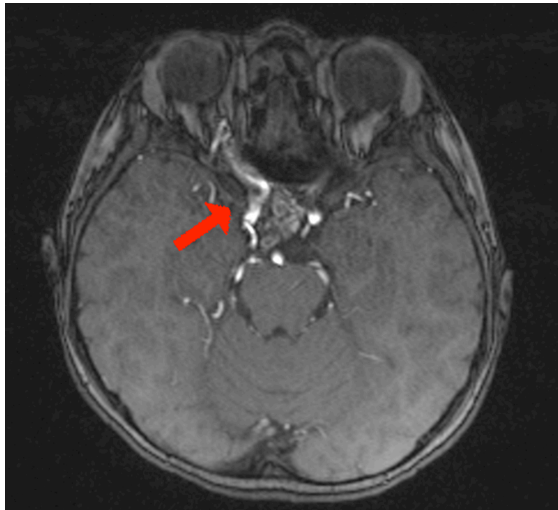


Figure 2A. The axial view of brain MRI showing the AVM located at the basal cistern and extending to the right third paraventricular (red arrow)

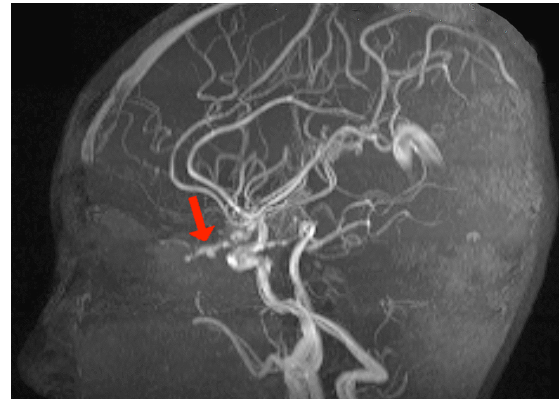


Figure 2B. The axial view of brain MRA showed the extended AVM at the retrobulbar area of the right eye (red arrow).

DISCUSSION

Wyburn-Mason syndrome is one of the rarest types of phacomatosis, a group of syndromes characterized by hamartomas of the eye, brain, skin, and occasionally the viscera and bones. In contrast to the other types of phacomatosis, Wyburn-Mason syndrome rarely involved the skin, but it regularly presented with hemangiomas in the midbrain and ipsilateral retina.^[1]

The correlation between the retinal and midbrain vessel was related to the embryological relationship between those two areas. The developing optic cup and the anterior neural tube came from the same origin, the primitive vascular mesoderm. When an abnormality occurs during the developmental process of the primitive vascular mesoderm, the anterior plexus, which gives rise to the retina vessel and hyaloid, and the midbrain vessels, will be affected. On the seventh week of gestation, the optic cup will fuse, a disruption of the developing tissue after this week is less likely to affect both the retina and the brain.^[5,6] In this case, the disruption most probably occurred before the seventh week of gestation, thus gave rise to the arteriovenous malformation (AVMs) in the retina and midbrain.

There are numerous ocular manifestations related to the Wyburn-Mason syndrome, while most of it gives only a mild disturbance to the visual

acuity, there are some others that can give a serious deterioration the patient's visual acuity. Based on the Archer classification, the higher the classification the higher the risk for visual loss and intracranial arteriovenous malformations. Vitreous hemorrhage, retinal detachment, cystic retinal degeneration, ischemia, and infarction are some of the reasons behind the prominent visual loss. In addition, a damage to the structures adjacent to the intracranial AVMs, happened due to compression or infarction, can lead to a visual loss or visual field loss.^[3,6] This child loss her right vision might be due to compressive optic neuropathy by the AVMs that has reached the retrobulbar area.

Quite similar to ocular manifestations, most of the intracranial AVMs are asymptomatic throughout the patient's life.^[5] Our case showed that she never experienced any neurologic symptoms, such as headaches, seizures, and paralysis. However, spontaneous intracranial hemorrhage from the AVMs may occur, and it is far more frequently than intraocular hemorrhage. In this particular situation, the patient may have neurologic symptoms, markedly paralysis and focal neurologic deficits.^[1]

A thorough examination of the brain and skin is warranted to every patient presented with retinal AVMs. CT-scan or MRI can be a helpful tool to examine the extension and precise location of the AVMs affecting the brain. In addition, imaging can also provide information about the adjacent structure and its relation to the AVMs. Considered as a more superior tool to show the hemodynamics and arterial supply to the AVMs, cerebral angiography is reserved for the symptomatic cases because of the significant risk related to the procedure.^[5] We decided not only to perform the MRI but also the MRA, a type of cerebral angiography, because she had a significant ocular symptom which is no perception of

light.

There is no significant treatment regarding the cutaneous and orbital AVMs. Nevertheless, close follow-up needs to be made to monitor vascular complications and glaucoma. If there is a complication, anti-vascular endothelial growth factor, pan-retinal photocoagulation, or other treatments related to the symptom are needed. Surgical intervention for the intracranial AVMs is not mandatory due to the high risk of postoperative neurologic and ophthalmic sequelae. Furthermore, non-surgical intervention, such as embolization, also rarely been done, as lesions are typically stable. However, if there is a high risk of hemorrhagic, embolization may be needed.^[1,5]

CONCLUSION

There are various presentations of Wyburn-Mason syndrome, which is related to the location and extension of the AVMs. A thorough examination of the eye and brain is mandatory in every case. In addition, a close follow-up is needed to examine the possible complications that lead to visual loss. Thus, prompt treatment might be done to save their vision.

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