Child with Wyburn-Mason syndrome presenting with sudden onset of intracranial haemorrhage

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DESCRIPTION

A 10-year-old child presented to the paediatric emergency department with a history of two episodes of generalised seizure lasting 2–3 min each in the past 24 hours. Prior to seizure episodes, the patient had history of headache for 1 day and two episodes of projectile non-bilious vomiting. No other significant systemic complaints or history. The patient had normal developmental history and was immunised up to date. On general examination, the vitals were pulse: 78 bpm, respiratory rate: 22 breaths/min and blood pressure: 104/60 mm Hg; the patient had a low Glasgow Coma Scale (GCS) of 9/15 (E2M5V2) with

Figure 1 (A, B) Non-contrast CT of the brain demonstrating right gangliocapsular bleed with intraventricular extension (white arrows). (C, D) Axial CT angiogram images demonstrating arteriovenous malformations (AVMs) with aneurysm of draining vein (solid arrow), dilated internal cerebral vein and feeding vessel from right Posterior Cerebral Artery (white arrows). (E) Coronal CT angiogram section demonstrating AVMs (solid arrow) with draining vein (white arrow). (F) Sagittal CT angiogram image demonstrating aneurysm of the draining vein (white arrow).

Figure 2 (A, B) Axial CT angiogram demonstrating dilated superior ophthalmic vein (white arrow) and arteriovenous malformations (AVMs) adjacent to the optic nerve (solid arrow). (C, D) Coronal CT angiogram section demonstrates multiple AVMs (solid arrow) adjacent to the optic nerve (white arrow). (E, F) Sagittal CT angiogram images demonstrating multiple AVMs adjacent to optic nerve (solid arrow) and intracranial AVMs (white arrow).

unequal pupils, which showed sluggish reactivity. On Central Nervous System (CNS) examination, the patient had brisk bilateral lower limb reflexes with normal tone, rest of the examination could not be performed due to low GCS. Rest of the systemic examination was normal. The routine blood investigations were within normal limits.

Non-contrast CT of the brain revealed foci of parenchymal haemorrhage in right gangliocapsular region with perifocal oedema ([figure 1A,B](#)). The patient underwent CT angiography, which revealed intracranial and orbital arteriovenous malformations (AVMs) ([figure 1C–F](#)). The intracranial malformation was in the right gangliocapsular region with feeding vessels arising from right Posterior Cerebral Artery (PCA) and draining into

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Images in...

internal cerebral vein, two aneurysms were present in the draining vein. The right orbit showed features of AVMs in the retrobulbar region and along the optic nerve (figure 2A–F). Because of intracranial and orbital AVMs, the patient was diagnosed with Wyburn-Mason syndrome. On fundoscopic examination, multiple retinal angiomas were noted in the right eye.

Wyburn-Mason syndrome is a rare type of phakomatosis caused by developmental abnormality affecting primitive vascular mesoderm shared by the developing optic cup and anterior neural tube. The condition presents with unilateral vascular abnormalities involving the facial structure, orbits and brain; bilateral involvement has also been reported.^{1–3} These patients express an anomalous vessel consisting of arteries and veins with no capillary bed causing direct communication between the two.⁴ The patient can present with visual symptoms like monocular amblyopia, esotropia or both.⁵ The extent of the visual symptoms depends on the size, extent and location of the retinal AVM; they can even present with vision loss due to vitreous, intraretinal or macular haemorrhage, and neovascular glaucoma.⁶ The neurological manifestations of Wyburn-Mason syndrome are hemiparesis and haemorrhage; it is different from neurological manifestation of cerebral AVM, which presents with epilepsy.^{5–7} Cutaneous manifestation is seen in a minority of patients, which present as angiomas of the face, cheek, nose, mandible, palate, pharynx, maxilla and buccal mucosa.⁵

Patients are usually conservatively managed with close observation for changes in lesion size; surgical intervention has been successful for small AVMs and symptomatic haemorrhage.^{8–9} Surgical intervention of suprasellar AVM is frequently associated with visual loss.⁴ Non-surgical interventions are radiotherapy and embolisation. Radiotherapy is associated with

endocrine abnormalities affecting the hypothalamic–pituitary axis and embolisation is used for cases that present with haemorrhage and not used prophylactically since the AVMs are relatively stable.^{10–12} Complete neurological evaluation with MRI of the brain should be considered in cases with retinal AVMs.

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Learning points

- ▶ Wyburn-Mason syndrome presents with neurological, ophthalmic and cutaneous manifestations. Patients can present with either visual or neurological symptoms at initial presentation.
- ▶ Intracranial arteriovenous malformations (AVMs) are relatively stable, and embolisation is indicated in patients with intracranial haemorrhage.
- ▶ Any patient with retinal AVM should undergo neurological evaluation.

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