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 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 the inferior petrosal sinus. The arteriovenous malformation in the orbit was multiple in number. The patient underwent embolisation of the arteriovenous malformations in the orbit and the gangliocapsular region with Onyx (figure 2A,B). The patient had no further episodes of seizures after the embolisation. The patient was discharged on the 5th day and has been well on follow-up.

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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 diag-
nosed with Wyburn-Mason syndrome. On fundoscopic exam-
ination, multiple retinal angiomias were noted in the right eye.

Wyburn-Mason syndrome is a rare type of phakomatoses
cauised 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 commu-
nication 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.2,5,7 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 obser-
vation 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 asso-
ciated with visual loss.4 Non-surgical interventions are radio-
therapy and embolisation. Radiotherapy is associated with
endocrine abnormalities affecting the hypothalamic–pituitary
axis and embolisation is used for cases that present with haem-
orrhage and not used prophylactically since the AVMs are rela-
tively stable.10–12 Complete neurological evaluation with MRI
of the brain should be considered in cases with retinal AVMs.

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data and images, the compilation/editing of the subsequent report. DAMAR was
responsible for the initial image interpretation, with inputs from KS and J.J. Patient
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