Wyburn-Mason syndrome

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A 14-year-old girl presented with chronic headache and right-side visual difficulties. She had right facial pigmentation since birth. Ophthalmoscopy examination reveals multiple dilated vascular channels over the right retina suggestive of retinal arteriovenous malformation (AVM) (Fig. 1). Cerebral angiography reveals multiple cerebral AVMs on the ipsilateral side (Fig. 2). Features were in keeping with Wyburn-Mason syndrome (WMS).

WMS is a rare type of phakomatosis characterized by unilateral vascular abnormalities involving the facial structures, orbits and brain and most commonly associated with Wyburn-Mason, who initially categorized the entity with detailed descriptions of nine case histories [1]. Clinically, patients may present with cerebral or ocular symptoms.

Fig. 1 Ophthalmoscopy view

Fig. 2 Digital subtraction angiography (DSA)

Headaches, seizures or subarachnoid haemorrhage are the usual manifestations, likely caused by compression effect or rupture of AVMs into adjacent structures [2]. Ocular manifestations are usually an important clue to the diagnosis of this syndrome. The arterial blood supply is more often from the internal carotid system than from the vertebrobasilar or external carotid system. In our case, both systems were equally affected. Though retinal involvement is usual, it is not essential for the diagnosis of WMS.

References

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