WYBURN-MASON SYNDROME ASSOCIATED WITH MOYAMOYA DISEASE

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ABSTRACT

A report case of subcutaneous hemangiomas of the head and face, intraorbital hemangiomas, intracranial AVM and occlusion of distal left internal carotid artery was shown in a 17-year-old man. The association of the Wyburn-Mason Syndrome and Moyamoya disease in this patient was rare and was not seen in the literature. The imaging modalities were CT scan and angiography of the head.

INTRODUCTION

Wyburn-Mason syndrome is one of the rare group of disorders collectively known as phakomatoses. It consists of arteriovenous malformations affecting the visual pathways, the mid-brain and subcutaneous facial structures (1). Moyamoya disease is a kind of occlusive arteriopathy which is congenital or acquired (2). Cases of association between Wyburn-Mason syndrome and Moyamoya disease is considered rare. We report such a case by CT and angiographic imaging.

CASE REPORT

A 17-year-old woman developed right hemiparesis for 10 days. She was a known case of left orbital cavernous hemangioma and hemangioma at left temporal part of the soft tissue of the head, diagnosed 3 years ago. Thirty days prior to the admission she was infected by herpes zoster at her right part of the face. Afterwards, there was swelling of left eyelid, left exophthalmos and right hemiparesis. Blindness of left side was noted. Motor power of right extremities was grade III. Sensation of right side was impaired. The response of the Babinki's was abnormal.

Non contrast enhancement and contrast enhancement axial and coronal CT scan of the brain and orbits showed large calcified known hemangiomas at the subcutaneous part of the left side of the head and face and in the left orbital cavity. In the brain, a large nidus of arteriovenous malformation is noted at left splenium of corpus callosum and left deep parietal lobe. Left common carotid injection showed nearly complete occlusion of the distal internal carotid artery. Mild degree of collateral vessels or moyamoya vessels are noted at both left common and right internal carotid arterial injection. Due to this occlusion of left internal carotid artery, the demonstration of the intracranial AVM is not possible. (Fig. 1,2).

DISCUSSION

Phakomatoses is a group of disorders described by Van Der Hoeve in 1923. It includes angiomatosis retinae (Von Recklinghausen's disease), tuberous sclerosis (Bourneville's disease), encephalotrigeminal angiomatosis (Sturge-Weber disease), ataxia telangiectasis (Louis-Bar syndrome), and Wyburn-Mason syndrome (1).

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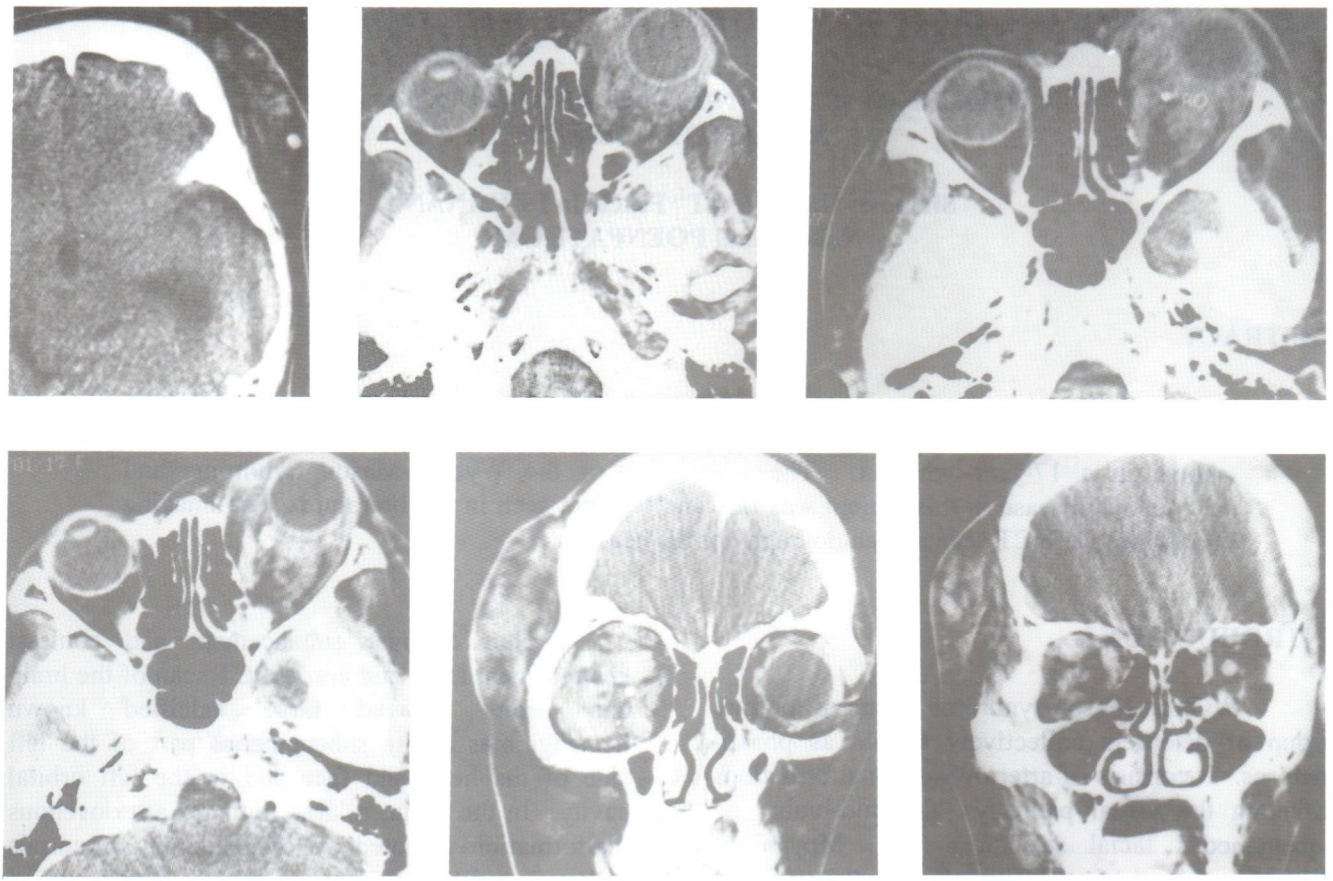

Fig. 1A. Axial nonenhanced CT scan of the orbits and enhanced coronal CT scan of the orbits showed the lesion of known hemangioma in the left orbital cavity, subcutaneous plane of left side of the head. Small calcification was noted in the lesions of both sites was shown.

Bonnet et al, first reported the association of retinal vascular malformations with ipsilateral cerebral arteriovenous malformations and facial naevi (3). The disorder stems from a vascular dysgenesis in the early embryonic period and extensions of the lesions vary widely. The first English report was by Wyburn-Mason who reviewed a large series in 1943 (4). The diagnosis was done only by other examinations than imaging.

The vascular abnormalities in the other phakomatoses are distinct from the arteriovenous malformations arising in Wyburn-Mason syndrome. In neurofibromatosis anomalies of the ophthalmic vessels and cavernous sinus are associated with sphenoid dysplasia. The vascular abnormality in Sturge-Weber syndrome is leptomeningeal angiodysplasia, with atrophy and calcification of the underlying cerebrum, Von Hippel-Lindau disease is an autosomal dominant angiodysplasia which consists of retinal capillary angioblastomas with multiple haemangioblastomas of the brain and spinal cord. Tumors affecting other viscera, especially the kidneys, are also a feature (1).

Manifestations of Wyburn-Mason syndrome can be cerebral or ocular or both. Headaches, seizures, or subarachnoid hemorrhage are the usual indicators of involvement of the central nervous system. The arterial blood supply is more often from the internal carotid system than from the vertebro-basilar or external carotid system. Venous drainage is primarily via the cavernous sinus or vein of Galen (1,5).

Damage to structures adjacent to the arteriovenous malformation can be due to compression from expansion of the vascular

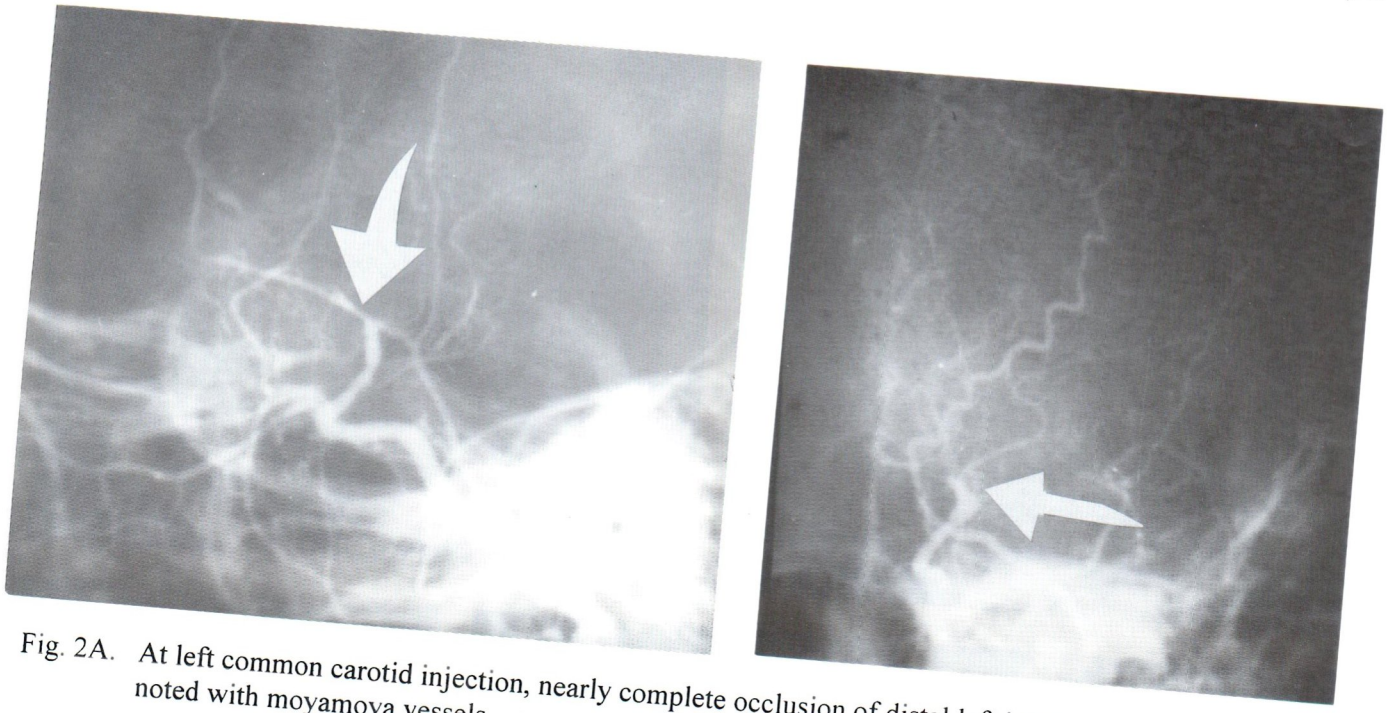

Fig. 2A. At left common carotid injection, nearly complete occlusion of distal left internal carotid artery was noted with moyamoya vessels.

malformation, hemorrhage into the adjacent structure, or due to ischemia from impaired perfusion (6).

Ocular manifestations are usually an important clue to the diagnosis of this syndrome. The retinal lesions, generally unilateral, range from ophthalmoscopically barely visible communications to large masses of tortuous and dilated vessels covering a substantial portion of the retina. The larger complexes usually cause cystic retinal degeneration between the dilated vessels and result in impaired vision (1,7). However, retinal involvement is not essential for the diagnosis of the Wyburn-Mason syndrome.

In the original series by Wyburn-Mason six patients had normal fundi. Other orbital manifestations which can occur include optic atrophy, enlargement of the optic foramen, and occasionally exophthalmos which may or may not be pulsatile (1,8).

Dermatologic lesions occur in a minority of cases and the clinical manifestation of these lesions varies from faint discolouration to extensive naevus involvement of the skin the trigeminal distribution, ipsilateral to the retinal or cerebral lesions. The deeper structures of the face such as the frontal and maxillary sinuses and the mandible can be involved (5).

Although arteriovenous malformations have little neoplastic potential they are dynamic structures and change with time. Substantial remodelling of an arteriovenous malformation has been observed in the retina and is evident on pathologic evaluation of intracranial lesions. Atrophy of some vessels occurs concurrently with dilation and expansion of others within the same lesion. These changes are most likely to be directed by haemodynamic factors caused by spontaneous thrombosis in parts of the complex. Progression of neurological signs can occur secondary to these changes (1,9).

Cases of association between these congenital disease and Moyamoya disease have been reported (10,11). Many other congenital diseases, such as von Recklinghausen's disease, tuberous sclerosis, Fanconi's anemia, and sickle cell anemia, are sometimes linked with a kind of occlusive arteriopathy termed Moyamoya syndrome (2,12-15). This seems to be the first report, demonstration of Wyburn Mason syndrome with associated Moyamoya disease.

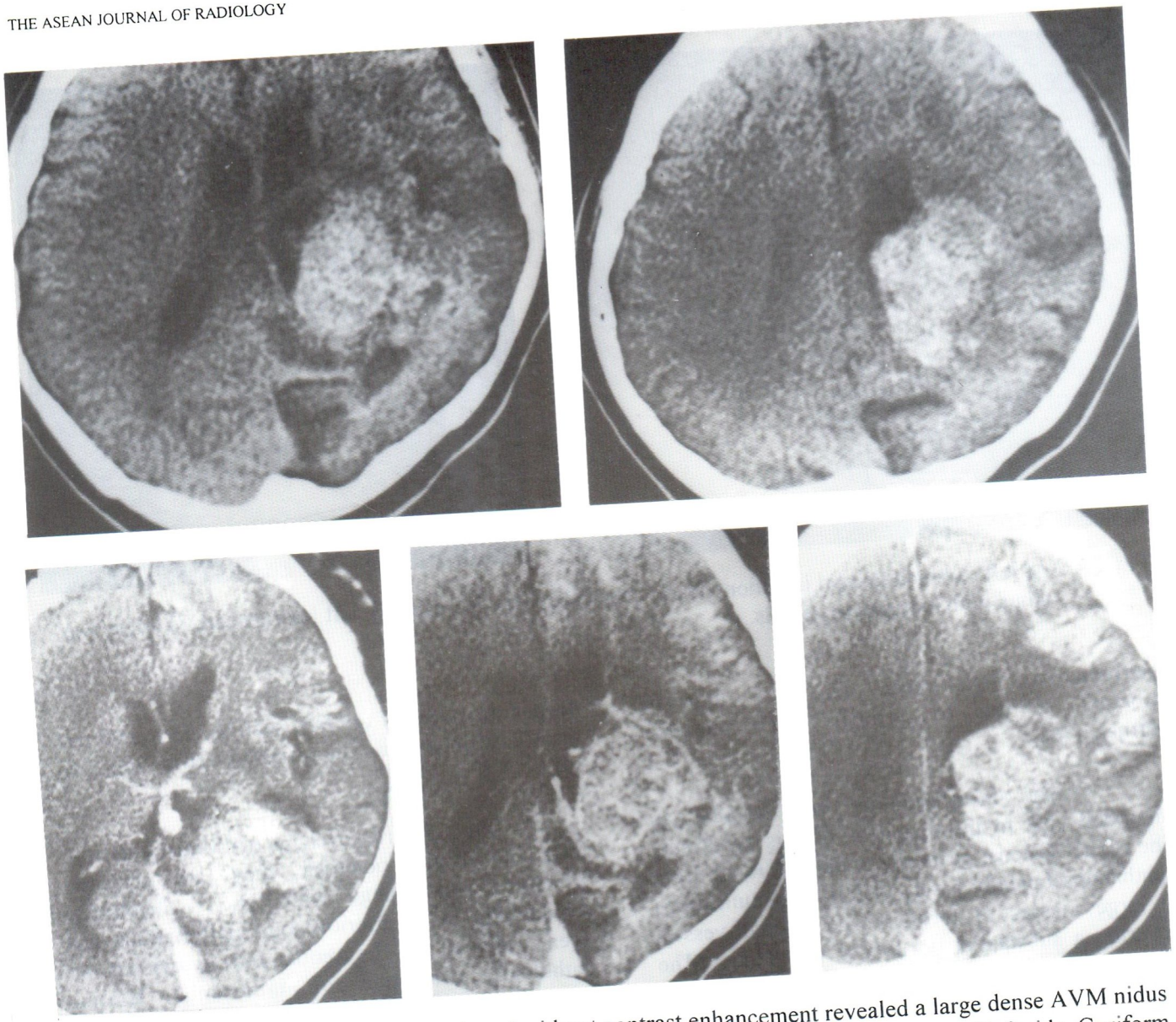

Fig. 1B. Axial CT scan of the brain with and without contrast enhancement revealed a large dense AVM nidus in the region of left splenium of corpus callosum, and around left posterior horn of left side. Gyriform enhancement of subacute left hemispheric ischemia was noted.

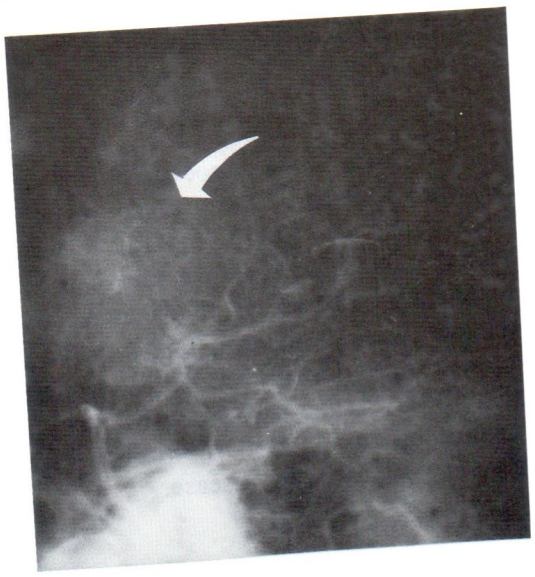

Fig.2B. At left vertebral injection, partial obstruction of left posterior cerebral and left cerebellar artery was noted. Moyamoya vessels are also noted from this injection.

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