Moyamoya:
Rare cause of stroke in a young patient

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Abstract

Moyamoya is a rare idiopathic progressive vaso-occlusive disease that involves the distal internal carotid arteries (ICAs) and circle of Wills. We present the case of a 40 year old female, Māori Etnicity, who presented with symptoms suggestive of a stroke, on the background of previous TIA like episodes in the past. Initial MRI brain was misleading,demonstrating an irregular pattern to the intracranial arteries and ICAs, giving rise to a suspicion of a vasculitis. However, digital subtraction angiography (DSA) showed a pattern of classic MMD.

Case Description

A 40 year old female presented to the Emergency Department with a 24 hour history of slurred speech, right sided upper limb weakness, and right sided facial droop. CT Head showed no evidence of an infarct or haemorrhage. She was commenced on treatment for an ischemic stroke, with a view to performing a MRI brain. However she discharged against medical advice, before this was performed. Three days later, the patient re- presented with right upper and lower limb weakness, aphasia, dysphagia and drooling. Neurological examination revealed the following findings: right sided facial droop, right sided upper motor neuron facial weakness, expressive dysphasia, homonymous hemianopia, a right sided hemiparesis (more pronounced in the right upper limb), and reduced sensation on the right hand side. MRI head showed large areas of restricted diffusion in the white matter of the left cerebrum, and the left frontal cortex, suggestive of a left MCA occlusion (Fig 1-2-3-4). In addition, an irregular pattern of the internal carotid artery and intracranial arteries pointed towards a possible vasculitic process as an etiology. On discussion with the Neurosurgical department in Auckland City Hospital, it was decided to transfer the patient to this centre for further investigation of a possible vasculitis. While the vasculitic screen was negative, a DSA revealed the following findings (Fig 5-6-7-8):

The supraclinoid portion of the right and left ICA, right M1, and proximal right and A1 were all narrowed, while the left M1 was occluded. Early basal ganglia collaterals were seen. These features were in keeping with a diagnosis of Moyamoya disease. Dual antiplatelet therapy was commenced for three weeks with a view to continuing clopidogrel lifelong. Unfortunately the patient discharged against medical advice one week later.

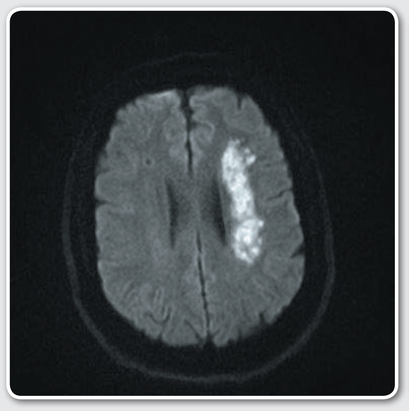


Fig 1: B1000 MRI Brain

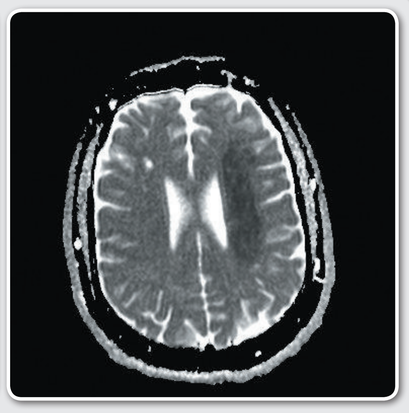


Fig 2: MRI brain ep 2d_diff _trace 55.0mm-6.5mm



Fig 3: MRA Brain

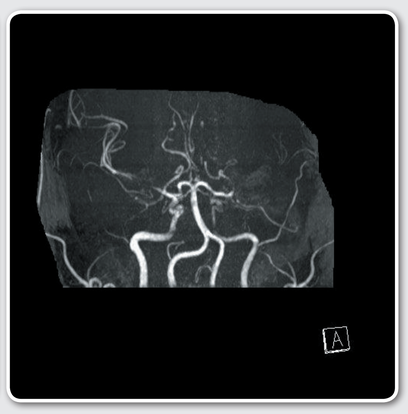


Fig 4: MRA Brain F-H cow

MRA Brain Fig 1,2,3,4: Conclusion: Large region of restricted diffusion in the left cerebral white matter with smaller areas of restricted diffusion in the left frontal cortex are likely acute infarcts as a result of occluded left middle cerebral artery. Intravenous gadolinium enhanced MRI is suggested to exclude tumour and infection. Irregularity of internal carotid and intracranial arteries suggestive of a vasculitis. Right white matter lesions are likely small vessel ischaemic disease with lacunar infarct.

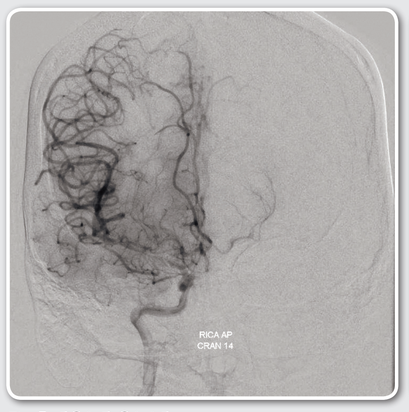


Fig 5: DSA RICA AP

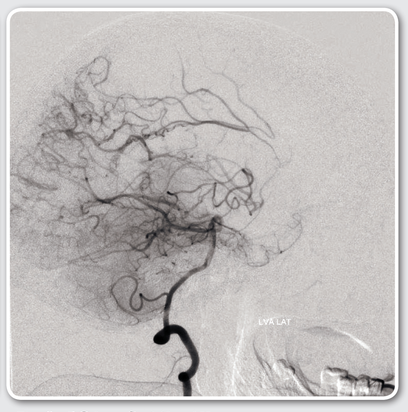


Fig 6: DSA Left LVA LAT

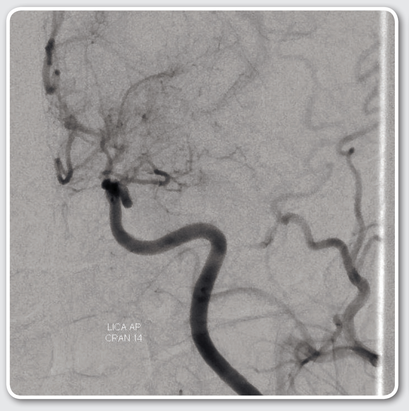


Fig 7: DSA LICA AP

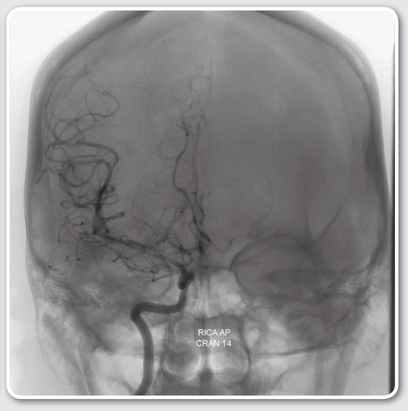


Fig 8: DSA RICA AP

DSA Fig 5,6.7.8:There is moderate narrowing of the supraclinoid right ICA, severe narrowing of the right M1 and critical narrowing of the proximal right A1. There is severe narrowing of the left supraclinoid ICA and A1 with occlusion of the left M1 and early basal ganglia collaterals. Vertebral angiography demonstrates much of both cerebral hemispheres to be supplied by parietal pial collaterals to the ACA and MCA.There is no significant large vessel atheroma. Occlusive intracranial arteriopathy with many features o classic moyamoya, though with poor basal ganglia collaterals at this point in time.

Discussion

Moyamoya disease is a chronic, progressive steno-occlusive disease affecting the terminal portions of the internal carotid arteries and the proximal portions of the anterior and middle cerebral arteries. (1, 2). As a compensatory mechanism to this stenosis, collateral vessels form, which are given the name “moyamoya vessels”(3, 4). The appearance of this abnormal vascular network, on angiography, has been likened to “something hazy, like a puff of cigarette smoke”, hence giving rise to the name Moyamoya, the Japanese term for a puff of smoke.(2). MMD has the highest incidence in Eastern Asia. Japan, for example has a prevalence of 6.03 per 100,000, and incidence of 0.54 per 100,000 per year, approximately 10 times that of Europe.(5) There is currently no data available in the literature of the frequency of Moyamoya disease in the Māori population, presumably due to the rarity of the disease. We suggest a national register of Moyamoya disease in this population, for future reference. MMD shows preponderance for female, at a ratio of 1.8. There are two peaks in life, in the 1st decade, most commonly between the ages of 5-10 years old, as well as the 3rd and 4th decades. (5) The pathogenesis has yet to be fully elucidated.(4) However a familial history in approximately 12.1% of MMD cases suggests the Importance of genetics in the process. (6). Genome wide linkage analyses have pointed towards an association between a missense variant of the ring finger protein 213 gene in chromosome 17q25.3, which increases susceptibility to MMD in East Asian populations/ those of East Asian ethnicity.(7, 8). MMD can also be seen in association with a number of haematological and autoimmune conditions, e.g. Graves disease, and type 1 diabetes, giving rise to the concept of the Moyamoya syndrome. (9-12). In terms of clinical manifestation, it can present in the form of cerebral ischemia (stroke or TIA), or haemorrhage. While most paediatric patients tend to develop ischemia, either can occur in the adult population.(13). Management can be divided into conservative and surgical. In terms of conservative management, although antiplatelet agents are widely used in non–Asian countries, evidence is currently lacking.(14). An interesting aspect of this case is the concomitant presence of schizoaffective disorder which preceded the diagnosis of Moyamoya disease by X years. There is limited literature on MMD in association with psychiatric disorders, and therefore conclusions cannot yet be drawn on whether a link, if any exists.

Case reports have described patients diagnosed with Moyamoya disease, and schizophrenia, psychosis, and depression. While there is not enough cases described as yet to be able to establish a link, if any, we recommend further research on the psychiatric history of patients diagnosed with MMD.

Conclusion

Moyamoya is a rare idiopathic progressive vaso-occlusive disease characterised by progressive steno-occlusive disease affecting the terminal portions of the internal carotid arteries and the proximal portions of the anterior and middle cerebral arteries. An MRA brain suggestive for vasculitis do not preclude the diagnosis and a digital subtraction angiography should be considered for young patients with stroke and abnormal MRA.

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