



Ocular Manifestation of Moyamoya Disease- Rare Case Report

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Abstract

Moyamoya disease (MMD) is a distinct entity, mostly found in Japan but cases are reported from many parts of the world. It is an angiopathy involving intracranial distal ends of internal carotid arteries (ICA). It may involve proximal parts of anterior cerebral artery and middle cerebral artery, causing narrowing of lumens resulting in ischaemia and collateral vessels formation. This leads to “puff of smoke” appearance, hence the name Moyamoya. MMD causes cerebrovascular ischaemic events, haemorrhagic strokes and seizures. Minor cases are observed and treated conservatively. Severe cases may need revascularization procedures. In our case report 8 years boy presented with loss of vision and optic atrophy in his right eye. Optic nerve involvement is rare but has been reported previously as well. In these cases monitoring of visual functions may need to done.

Keywords: *Moyamoya disease, Optic atrophy, cerebrovascular ischaemic event.*

Introduction

MMD was first described in Japanese in 1957^[1]. The name was coined in 1969 by Suzuki and Takaku which means “puff of smoke” in Japanese^[2]. This is owing to its angiographic appearance. This picture is seen because of a network of collateral vessels in the vicinity. MMD is an angiopathy with progressive narrowing, affecting distal intracranial parts of internal carotid arteries, as well as proximal parts of anterior and middle cerebral arteries. Posterior circulation is intact. The exact cause is unknown. Women are affected nearly twice as men^[3]. The condition is responsible for ischaemic cerebrovascular events and haemorrhagic strokes. Intra cranial haemorrhages are more common in

adults as compared to children^[4], some 20% of adult patients develop these episodes. Bleeds are mainly intraparenchymal and intraventricular due to rupture of fragile collateral blood vessels. The onset of MMD has two peaks, one in childhood between 5 to 8 years of age, another in adults in their fourth decade.^[5]

Apart from strokes other features of MMD include intellectual disability, seizures, and Migraine-like headaches. Intellectual disability is common in children who have had strokes^[6]. Cognitive functions are also affected probably due to chronic cerebral hypoxia^[7] Optic nerve involvement is very rare. Only 2 cases were reported earlier, so far^{[8][9]}.

Magnetic Resonance Imaging (MRI) and Magnetic Resonance Angiography (MRA) are the key investigations in diagnoses of MMA. These help in diagnosis and progress of disease.

Histopathologically there is concentric and eccentric fibrocellular thickening of the intima in the distal parts of ICA, leading to stenosis, and fragmentation of internal elastic lamina.^[10]

Case Report

This 8 years old male child initially presented to paediatric unit in a district hospital in Lancashire United Kingdom with right sided weakness and seizure. He was then referred to neurologist who diagnosed Moyamoya disease after MRI and MRA. Acquired brain injury due to MMD was noted. He had hypertension and learning difficulties. He was put on Aspirin and Amlodipine. Epilepsy was treated with anticonvulsants and surgical intervention was planned. A few months later his loss of vision in right eye was noted which triggered his referral to our Ophthalmology department.

He had bilateral terminal internal carotid stenosis on angiogram which appeared worse on the right side but he was symptomatic on left, despite angiographic findings. His left hemisphere and most of the right hemisphere was filled from right internal carotid. Other features of MMD were also present. He had left middle cerebral artery infarct. He was seen by multidisciplinary team throughout.

His visual acuity was markedly reduced in right eye with a best correct visual acuity (BCVA) of counting fingers (CF). On examination; he had right relative afferent pupillary defect (RAPD) and atrophic optic disc in the same eye. Apart from that eye examination was normal. The left eye had perfectly normal vision. Slit lamp examination and fundoscopic examination was normal. Orthoptically he was stable. Visual field test 120 points screening was done which was full in his left eye. He was followed up in eye clinics with Visual fields on each visit to observe. His left eye

remain normal throughout. He later developed divergent squint in the right eye.

Patient had bilateral Pial Synangiosis to improve his circulation a few months later, with good results. He is still under the neurologist with no symptoms suggestive of ischaemia. He also has periodic ophthalmology review with visual field for his left eye for observation.

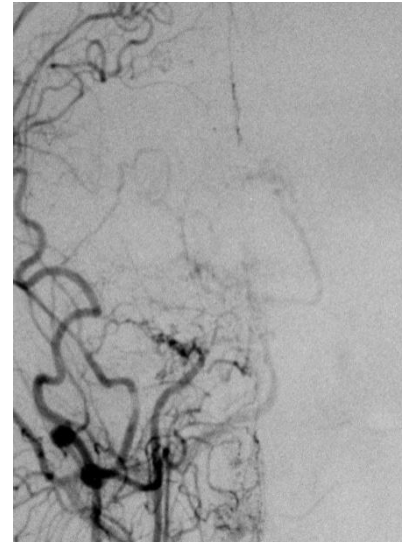


Figure 1: MRA showing “puff of smoke” appearance

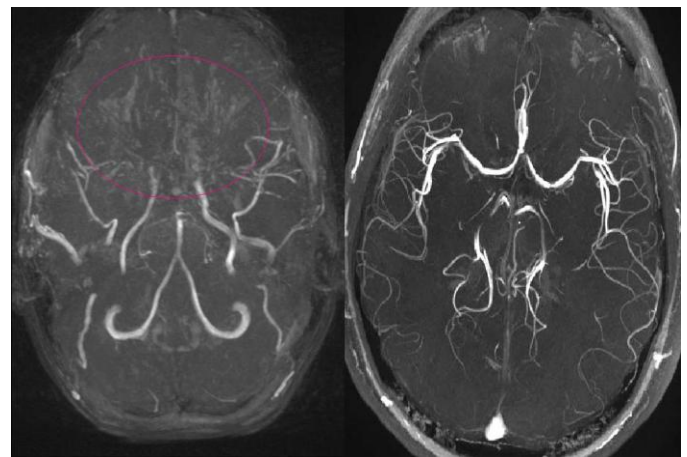


Figure 2: Right-MRA for MMD in patient, Left-normal patient for comparison

Dicussion

Moyamoya disease has been reported in east and west but is seen to be more common in Japan and Korean peninsula. Asymptomatic cases have also been noted. These cases and those with milder symptoms are treated conservatively with antiplatelets and antifibrinolytics. Epilepsy is

managed by anticonvulsants. Severe cases are treated surgically by revascularization which may be direct or indirect bypass or a combination of the two.

In our case patient was already diagnosed by neurologists and neurosurgeons. He was referred for visual deterioration in right eye. Patient had already lost vision to CF in this eye and the optic disc was atrophic. It was unilateral case of optic atrophy. Visual acuity and visual field in left eye was perfectly normal and remained so subsequently. Patients still has follow up appointments in our Ophthalmology department with Visual fields to strictly observe the other eye. Optic nerve is composed of axons of retinal ganglion cells. Any insult to these axons results in optic atrophy. Optic atrophy and loss of vision in right eye in these cases is attributed to ischaemia due to involvement of short posterior ciliary arteries. It can also happen due to involvement of central retinal artery but in our case retinal vessels on fundus examination were normal without any tell-tale signs of previous retinal artery occlusion.

Disclosure: The author declares that he has no conflict of interest.

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