



Case report

MOYAMOYA DISEASE - CLINICAL PROGRESSION AND LONG-TERM OUTCOME: A CASE REPORT

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ABSTRACT

Moyamoya disease (MMD) is a chronic cerebrovascular disease, characterized by stenotic- obstructive changes at the terminal portions of the internal carotid artery (ICA) and formation of an abnormal vascular network at the cerebral base. The clinical presentation of MMD may vary from TIA and ischemic or hemorrhagic strokes to seizures, headaches and cognitive impairment. Typically strokes in the MCA and ACA are prevalent at the initial stages, while PCA vascular incidents increase with disease progression and are considered to be a poor prognostic factor.

We present a case of a 21-year-old man admitted at the Neurology Department with Jacksonian seizures in the right arm followed by right-sided weakness and sensory-motor aphasia. Over the following decades he suffered subsequent transitory episodes of visual impairment, one ischemic stroke with right-sided weakness and a left thalamic hemorrhage. CT scans, angiographies and MRI showed multiple ischemic lesions. A catheter-based DCA showed multiple stenotic regions in the ICA, MCA and ACA and occlusion of the left PCA in P1 and well-developed collateral network of the right PCA thus confirming the diagnosis of MMD.

Moyamoya should always be considered as a possible cause for vascular events in young adults and especially in children. Therapeutic measures are focused on secondary prophylaxis of ischemic events with antiplatelets as there is no specific treatment available to stop the progression of the disease. Current guidelines suggest surgical methods as the only effective prevention of further disabling ischemic and hemorrhagic strokes.

Keywords: moyamoya disease, ischemic stroke, hemorrhagic stroke,

INTRODUCTION

Moyamoya disease (MMD) is a chronic cerebrovascular disease, characterized by stenotic- obstructive changes at the terminal portions of the internal carotid artery (ICA) and formation of an abnormal vascular network at the cerebral base. [1, 2] It presents as either unilateral or bilateral ICA terminal stenosis and could be an important cause of non-atherosclerotic intracranial disease, particularly in East Asian countries where the condition is significantly more prevalent. [2, 3] Two peaks of disease incidence are reported – at around 10 years and at 30 years, both with different clinical presentations. Pediatric cases present more commonly with ischemic events, while young adult patients usually present with brain hemorrhages. MMD affects predominantly the anterior circulation, usually originating from the distal portions of the ICA and the proximal portions of the MCA and ACA. Brain hypoperfusion leads to recruitment of collateral vessels from the leptomeninges and eventually from anastomotic branches of the ECA (external carotid artery). Bilateral involvement is defined as moyamoya disease (MMD), while unilateral is usually referred to as moyamoya syndrome (MMS) and is considered to be caused by other pathological processes. [4] The pathophysiology of MMD is not completely understood. Studies demonstrate that common pathologies like atherosclerosis or inflammation are not the cause for the stenosis in MMD. Histopathologic specimens show proliferation of the smooth muscle cells, irregular layering of the elastic lamina and micro-aneurysm formation. Evidence supports the theory that apoptosis and not necrosis is the leading cause for the tunica media changes, probably mediated by caspase mechanism triggered by an underlying autoimmune process – elevated antibodies have been discovered as wells as higher plasma concentrations of angiogenetic factors like VEGF (vascular endothelial growth factor), MMP (matrix metalloproteinase) and interleukin-1B. [5] As mentioned above hypoperfusion of the brain leads to compensatory enlargement and dilation of small perforator branches causing fragmentation of the elastic layer and making the walls weaker and prone to aneurysmal formation and potential rupture. [6] The clinical presentation of MMD may vary from TIA and ischemic or hemorrhagic strokes to seizures, headaches and cognitive impairment. The most common event at presentation

is ischemic stroke or TIA due to the cerebral hypoperfusion. [7, 8] Typically, TIAs can be triggered by hyperventilation, possibly due to vasodilation of normal vessels induced by the decrease in PaCO₂ and subsequent hypoperfusion of affected areas via a steal phenomenon. [1] Although strokes in the MCA and ACA are prevalent at the beginning stages, PCA involvement and vascular incidents increase in prevalence with the disease progression and are considered to be a poor prognostic factor. Hemorrhagic strokes are regarded as a complication of the later stages of disease progression. [7, 8, 9] Studies show that intraventricular hemorrhages occur much more frequently in MMD than in primary hypertensive hemorrhage. [10] The gold standard for diagnosis of MMD is catheter-based DCA and it is also crucial for the selection of the ECA branch most suitable for bypass. MRI imaging shows areas affected by acute ischemic strokes, while perfusion-weighted imaging (PWI) and CT perfusion are used for pre- and postoperative evaluation of patients. No effective treatment is available to stop the natural progression of the disease. Therapies are based on secondary prophylaxis with antiplatelet drugs and surgical revascularization to restore blood flow of the hypoperfusion areas.

CASE REPORT

A 21-year-old man was admitted at the Neurology Department of UMHAT-Pleven in 1989 with Jacksonian seizures in the right arm and subsequent right-sided weakness and sensory-motor aphasia preceded by a febrile state of viral etiology. CT showed several ischemic zones in the left hemisphere, while SPECT revealed several hypoperfusion areas – in the left frontal, right temporal and both occipital lobes. Initial sonography of the cerebral vessels was unremarkable, while the one done several years later showed changes in the blood flow of both MCAs and collateral blood supply via the two posterior communicating arteries. CSF testing was normal, as well as the CSF electrophoresis. The patient was treated and discharged with recommendations for secondary prophylaxis with ASA. In the following years after his initial presentation the patient complained of multiple transitory episodes of visual impairment – blurred vision, scotoma of the left eye, and one further ischemic stroke with right-sided hemiparesis. In 2008 the patient was diagnosed with paroxysmal atrial fibrillation. In 2022, 33 years after the initial vascular incident, the patient presented with acute headache, right-sided hypoesthesia, visual and speech impairment and increased blood pressure. CT scan showed blood collections in the left thalamus and in the basal cisternae, while CT angiography was suggestive of an aneurysm of the left PCA. MRI showed multi-infarct encephalopathy, stenotic changes in both ACAs, the right MCA and the left ICA, but did not visualize the aforementioned aneurysm of PCA. In further testing a catheter-based DSA was performed and confirmed multiple stenotic regions in both ICA, the right MCA and ACA, occlusion of the left PCA in P1 and well-developed collateral network of the right PCA. The DSA findings officially confirmed the diagnosis of MMD.

Fig. 1. DSA showing stenosis of ICA



Fig. 2. DSA showing multiple stenotic regions



DISCUSSION

Our patient presented with first ischemic stroke at an age between the two peak ages for MMD, although possible TIAs may have been present before the initial stroke. The disease progression was typical for MMD – with multiple ischemic incidents and TIAs, Jacksonian seizures and hemorrhagic stroke both in the brain parenchyma and in the subarachnoid space three decades later. With the disease progression he developed multi-infarct encephalopathy with cognitive impairment. Neuroimaging showed multiple stenotic changes initially of the anterior circulation vessels that later on involved also the posterior circulation and formation of an abnormal network of collateral vessels.

CONCLUSION

Moyamoya is a chronic progressive disease that affects younger patients and leads to irreversible and potentially fatal complications. Although rare, it should always be considered in case of vascular events in young adults and especially in children. Therapeutic options are limited and aimed predominantly at secondary prophylaxis with antiplatelet drugs. Surgical options should be strongly considered especially in the beginning stages of the disease, as direct and indirect bypass procedures that anastomose intra- and extracranial vessels have proven to be effective for prevention of further strokes and progression of the disease.

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