

MOYAMOYA DISEASE

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Abstract: Moyamoya disease (MMD) is a cerebrovascular disease of unknown etiology, mainly reported in Japanese. It is characterised by typical angiographic findings of bilateral occlusion at the terminal branches of the internal carotid artery together with network of collaterals at the base of the brain resembling a "puff of smoke". There is evidence to show linkage of moyamoya to several chromosomes. A role of various cytokines, growth factors, prostaglandins and cerebrospinal fluid proteins in its pathogenesis has been proposed and the role played by infections remains to be elucidated. Better methods for studying the cerebral blood flow (CBF) and screening have been devised. Various surgical procedures have been developed for revascularization.

INTRODUCTION

MMD is a clinical entity of unknown etiology, probably an inherited vasoocclusive disease. It was first reported in Japanese. The term moyamoya means "a wavering puff of smoke drifting in the air". It is characterised by pathognomic angiographic findings of bilateral occlusion at the terminal portion of the internal carotid artery together with abnormal vascular network at the base of the brain.

EPIDEMIOLOGY & ETIOLOGY

MMD is prevalent in Japan and Asian countries¹. A genetic mode of inheritance with familial occurrence has been suggested². Various cytokines have been shown to be related to the pathologic thickening of the intima of cerebral vessels³. The role of prostaglandins in its pathogenesis has been studied⁴. Elevation of nitric oxide metabolite⁵ and detection of some specific polypeptides in CSF⁶ have been reported to be specific for MMD. Role of infection particularly by cytomegalovirus and Epstein Barr virus has been suggested⁷.

Staging of MMD⁸ is done according to MR angiography (MRA) *Stage I:* narrowing of aortic fork; *Stage II:* initiation of moyamoya; *Stage III:* Intensification of moyamoya; *Stage IV:* Minimization of moyamoya; *Stage V:* Reduction of moyamoya; *Stage VI:* Disappearance of moyamoya.

CLINICAL FEATURES

MMD is common in children, recurrence is a characteristic feature. Children present with transient ischemic attacks, reversible ischaemic neurological deficit, ischaemic strokes, hemiplegia, hemiparesis, monoparesis, sensory impairment, headache, dizziness, seizures, involuntary movements of limbs or mental retardation. Adults usually present with sudden onset intracranial hemorrhage in the form of intraventricular, subarachnoid or intracerebral type^{1,8}. Skin lesions in the form of livedo reticularis have been described (*Sneddon syndrome*⁹). Association with malformations eye i.e. morning glory disc and chorio-retinal- colobomas have also been reported¹⁰; frequently associated with the base of skull malformations like basal meningoencephalocele, panhypopituitarism and midface malformations¹¹. The natural history of MMD is not yet definitely known; 75 to 80% of cases tend to have a benign course with regard to life expectancy with or without surgical treatment.

NEUROIMAGING

- Cerebral angiography:** It is the *gold standard* for diagnosing the disease and is indispensable for carrying out revascularization procedures; cerebral aneurysms are found in 10% of cases¹².
- CT Scan:** 40% of cases with cerebral ischaemia show low density areas in the cerebral cortex and or subcortical white matter, cerebral atrophy, dilatation of third and lateral ventricles. Cases with haemorrhage show high density areas corresponding to bleeding in the basal ganglia, thalamus, ventricle, subcortex and

cortex.

- MRI:** It is not only pathognomic but also detects small infarcts, hemorrhage and abnormal collaterals.
- EEG:** EEG recordings include posterior slowing and centrottemporal slow wave activity and prolonged hyperventilation.
- Position emission tomography (PET):** It reveals reduction of cerebral blood flow (CBF), reduced regional cerebral metabolic rate of oxygen and increase in regional oxygen extraction fraction in the cerebral hemisphere except in the basal ganglia.

Cerebral blood flow measurement: CBF can be measured by Xenon-enhanced CT, single photon emission computed tomography or PET. MMD is associated with a diffuse reduction of CBF especially in the frontal lobes. But CBF in the basal ganglia is preserved and it decreases with the advancing age¹³.

TREATMENT

There is no specific treatment. **Medical therapy** includes use of vasodilators, anticoagulants (ASA), low molecular weight dextran, steroids and anticonvulsants but then usually with little effect. **Surgical treatment** includes various revascularization procedures in the form of:

- Direct revascularization procedures**¹⁵ i.e. superficial temporal middle cerebral artery bypass and extracranial intracranial bypass to anterior or posterior cerebral artery.
- Indirect revascularization procedures**¹⁶ i.e. encephalo-duro-arterio-synangiosis (EDAS), encephalo-myo-synangiosis (EMS), pial synangiosis, encephalo-arterio-synangiosis (EAS), intracranial omental transplantation and muscle transplantation (latissimus dorsi, gracilis). Generally indirect revascularization is preferred in children.

REFERENCES

- Hoffman HJ. Moyamoya disease and syndrome. *Clin Neurol Neurosurg* 1997; 99 (Suppl 2): 539-44.
- Shetty AN, Alva S. Familial moyamoya disease in caucasians. *Pediatr Neurol* 2000; 23: 445-47.
- Aoyagi M, Fukai N, Yamamoto M et al. Early development of intimal thickening in superficial temporal arteries in patients with moyamoya disease. *Stroke* 1996; 27: 1750-54.
- Yonekawa Y, Ogata N, Kaku Y et al. Moyamoya disease in Europe, past and present status. *Clin Neurol Neurosurg* 1997; 99 (Suppl 2): 558-60.
- Noda A, Suzuki M, Takayasu K et al. Elevation of nitric oxide metabolites in cerebrospinal fluid of patients with moyamoya disease. *Acta Neurochir (Wien)* 2000; 142: 1275-80.
- Hoji M, Hoshimaru M, Miyamoto S et al. Cerebrospinal fluid proteins associated with moyamoya disease: report of three cases. *Neurosurg* 1999; 45: 170-74.
- Tanigawara T, Yamada H, Sakai N et al. Studies on cytomegalovirus and Epstein Barr virus infection in moyamoya disease. *Clin Neurol Neurosurg* 1997; 99 (Suppl 2): S 225-28.
- Suzuki J, Takasu A. Cerebrovascular moyamoya disease. Disease showing abnormal net like vessels in the base of brain. *Arch Neurol* 1969; 20: 288-99.
- Soti IA, Boyle RS. Sneddon's syndrome. *Aust NZJ Med* 1986; 16: 799-802.
- Krishnan C, Roy A, Traboulsi E. Morning glory disk anomaly, choroidal coloboma and congenital constrictive malformations of the internal carotid arteries (Moyamoya disease). *Ophthalmic Genet* 2000; 21: 21-24.
- Morniyama M, Yasui T, Sakamoto H et al. Basal meningoencephalocele, anomaly of optic disc and panhypopituitarism in association with Moyamoya disease. *Pediatr Neurosurg* 2000; 33: 100-104.
- Waga S, Tachio H. Intracranial aneurysm associated with moyamoya disease in childhood. *Surg Neurol* 1985; 23: 237-47.
- Handa J, Nakano Y, Okuno T et al. Computerized tomography in moyamoya syndrome. *Surg Neurol* 1977; 7: 315-19.
- Fujisawa I, Asato R, Nishimura K et al. Moyamoya disease: MR imaging. *Radiology* 1987; 164: 103-105.
- Iwama T, Hashimoto N, Miyake H et al. Direct revascularization to the cerebral artery territory in patients with moyamoya disease. Report of five cases. *Neurosurgery* 1998; 42: 1157-61.
- Mizoi K, Kayama T, Yoshimoto T et al. Indirect revascularization for moyamoya disease: Is there a beneficial effect for adult patients? *Surg Neurol* 1996; 45: 541-48.

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