International Journal of Contemporary Pediatrics
Bainade K et al. Int J Contemp Pediatr. 2025 Jul;12(7):1223-1225
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

Case Report

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20251879

Moyamoya disease: a silent smoke

Kapil Bainade, V. Kotrashetti, Neelu Elon, Saylee Jagtap, Yash D. Vasagadekar*

Department of Pediatrics, Dr. D. Y. Patil Hospital, Nerul, Navi-Mumbai, Maharashtra, India

Received: 11 April 2025
Revised: 17 May 2025
Accepted: 22 May 2025

***Correspondence:**
Dr. Yash D. Vasagadekar,
E-mail: yashvasagadekar5319@gmail.com

ABSTRACT

Moyamoya disease (MMD) is an isolated chronic, usually bilateral, vasculopathy of undetermined etiology characterized by progressive narrowing of the terminal intracranial portion of the internal carotid artery and circle of Willis. It is a connective tissue disorder of cerebrovascular vessels of unknown aetiology. A fragile network of abundant collateral vessels as a reaction to chronic brain ischemia develops predominantly at the base of the brain known as moyamoya vessels. This activity describes the pathophysiology, evaluation, and management of MMD and highlights the role of the interprofessional team in the management of affected patients. Here we present a case of 3-year-old female child who had frequent transient episodes of twitching of left side of her face with deviation of angle of mouth towards right side since 15 days without neurological deficit and altered sensorium. There also was a history of transient episode of left sided paresis at the age of 18 months of life. Detailed evaluation and magnetic resonance imaging (MRI) confirmed the diagnosis of MMD. In this particular case the age group of presentation for MMD is unusual. Discussion of this case will help to spread awareness about early suspicion and appropriate intervention at right time to improve the developmental and cognitive status of such cases.

Keywords: Moya Moya, Neurosurgery, Developmental status, Cognitive status, Surgical intervention

INTRODUCTION

Moyamoya disease (MMD) is idiopathic, meaning its cause is unknown. It features progressive narrowing or blockage of the supraclinoid segment of the internal carotid arteries (ICAs), leading to the formation of compensatory collateral vessels, which appear like a "puff of smoke" (the meaning of moyamoya in Japanese) on angiography.[1]

Moyamoya syndrome (MMS) involves the same vascular changes, but they occur secondary to an underlying condition, which could be: neurological (e.g., neurofibromatosis type 1), hematologic (e.g., sickle cell disease), autoimmune (e.g., systemic lupus erythematosus), and infectious, radiation-induced, or genetic syndromes.[1]

CASE REPORT

A 3-year-old FCH was brought with the complaints frequent episodes of twitching over left side of her face with deviation of angle of mouth towards right side, which lasted for 10-15 minutes, aborted on its own, without any neurological deficit or altered sensorium after event. At 18 months of age, patient had transient episode of weakness of left upper and lower limb followed by an episode of generalized tonic clonic convulsion. Birth history was uneventful and developmentally child was normal. She is fully immunized till date. Detailed evaluation and neuroimaging was done which was suggestive of acute infarct in right middle cerebral artery and watershed area, for which Aspirin and Levetiracetam were started. Further magnetic resonance imaging (MRI) brain angiography confirmed the diagnosis of MMD. Electroencephelo-

graphy (EEG) suggestive of abnormal epileptiform activity over both cerebral hemispheres. Computed tomography (CT) angiography was done by neurosurgeons to look for patency of blood vessels. Patient was taken up for intracranial bypass surgery. Post-surgery review CT brain was suggestive of improvement in radiological picture. Simultaneously clinical features also improved (twitching had stopped).

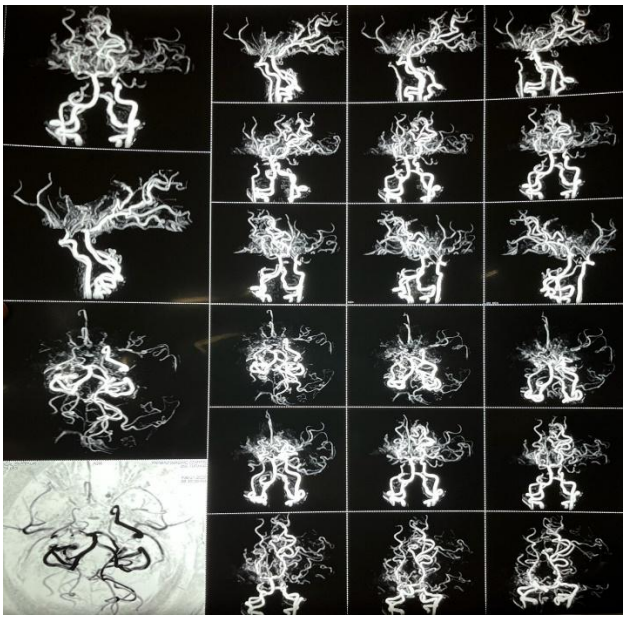

Figure 1: MR angiography brain suggestive of puff of smoke appearance (Moyamoya).

Figure 2: 3-year-old ready for surgery.

DISCUSSION

MMD is a rare, progressive cerebrovascular disorder affecting the carotid arteries, particularly their terminal (supraclinoid) segments. These arteries supply critical blood flow to the brain. The name "Moyamoya", which means "puff of smoke" in Japanese, describes the appearance of the fine collateral vessels that develop to compensate for the arterial stenosis, as seen on cerebral angiography. The disease was first described in Japan in the 1950s and was once considered primarily an East Asian condition. However, it has since been reported in diverse populations worldwide, including North America, Europe, and Africa.[2] In MMD, the internal carotid arteries (ICAs), which supply oxygen-rich blood to the brain, become progressively thickened and narrowed over time due to the occlusion of their terminal portions (the supraclinoid segment). As a result, the brain begins to experience a reduction in blood flow, leading to a state of ischemia (insufficient blood supply). To compensate for this loss of perfusion, the body forms collateral circulation—new, smaller arteries that attempt to bypass the blocked sections and provide alternative pathways for blood flow. These newly formed vessels, often referred to as Moyamoya vessels, are fragile and can be prone to rupture or thrombosis. While they attempt to compensate for the reduced blood flow, they may not always be able to supply sufficient oxygenated blood, which can lead to symptoms like stroke, transient ischemic attacks (TIAs), and neurological deficits.[2] While MMD is rare, its prevalence varies across different regions. In global studies, the disease prevalence has been reported to range from 3.2 to 10.5 cases per 100,000 populations. However, there are no large community-based studies specifically from India, making it difficult to ascertain its true prevalence in that region.[2] MMD is more prevalent among Asians, with approximately 57% of affected individuals coming from Asian countries, particularly Japan, Korea, and China. This geographic distribution suggests there may be genetic or environmental factors influencing the disease's occurrence. However, the exact cause of MMD remains unknown. It is considered an idiopathic condition, meaning it occurs without a known cause in the majority of cases.[2] Although the disease may be seen in any age group, it is more common seen in two distinct age groups; 5-15 years and 30-40 years of age.[2] The symptoms of MMD primarily arise due to two key factors: reduced blood supply to the brain, caused by the progressive narrowing or occlusion of the internal carotid arteries; and rupture of Moyamoya vessels, the fragile collateral vessels formed to compensate for the reduced blood flow.[2]

Children may have weakness or numbness of an arm or leg, hemiparesis, monoparesis, involuntary movements, headaches, dizziness, or seizures.[2] Mental retardation or persistent neurologic deficits may be present. Adults may have symptoms and signs similar to those in children, but haemorrhage (bleeding) of sudden onset is more common in adults.[2]

The condition is diagnosed on MRI and cerebral angiography in a patient presenting with stroke or intracranial bleed.[2]

MMD has been found to co-occur with several other neurological and systemic conditions. Though the exact cause-and-effect relationship remains unclear, the association between these conditions and MMD is well-documented. Some of the most common conditions that are seen alongside MMD include Down's syndrome,

neurofibromatosis type 1, sickle cell disease, congenital heart disease, and radiotherapy to head and neck.[2]

Since the exact cause of the disease is unknown, there is no treatment to reverse the narrowing of vessels. However, the treatment is aimed at preventing stroke by administering aspirin and surgery that improves blood flow to the affected parts of the brain. Existing studies have proved that antiplatelet therapy has long-term benefits for patients with MMD.[3] Surgery is highly successful in preventing stroke and bleeding in future and is the mainstay of treatment. Without surgery, the majority of individuals with paediatric MMD will experience mental decline and multiple strokes because of the progressive narrowing of arteries.[4] Surgical interventions (combination of indirect and direct revascularization techniques) was found to be more effective than conservative treatment in preventing poor cognitive outcome, especially if done early in the clinical course.[5]

CONCLUSION

In pediatric MMD, early suspicion and detection along with early surgical intervention should be advised so as to prevent cognitive decline. Surgical revascularization has been proven beneficial in slowing cognitive decline in paediatric moyamoya patients.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Scott RM, Smith ER. Moyamoya disease and moyamoya syndrome. N Engl J Med. 2009;360(12):1226-37.
2. Suzuki J, Takaku A. Cerebrovascular "Moyamoya" disease. Disease showing abnormal net-like vessels in base of brain. Arch Neurol. 1969;20(3):288-99.
3. He S, Zhou Z, Cheng MY, Hao X, Chiang T, Wang Y, et al. Advances in moyamoya disease: pathogenesis, diagnosis, and therapeutic interventions. MedComm (2020). 2025;6(2):e70054.
4. Guzman R, Lee M, Achrol A, Bell-Stephens T, Kelly M, Do HM, et al. Clinical outcome after 450 revascularization procedures for moyamoya disease. Clinical article. J Neurosurg. 2009;111(5):927-35.
5. Kuroda S, Houkin K. Moyamoya disease: current concepts and future perspectives. Lancet Neurol. 2008;7(11):1056-66.

Cite this article as: Bainade K, Kotrashetti V, Elon N, Jagtap S, Vasagadekar YD. Moyamoya disease: a silent smoke. Int J Contemp Pediatr 2025;12:1223-5.