# Moyamoya disease presenting as acute onset cortical blindness: a case report

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**Abstract**: We report a case where acute onset cortical blindness is the mode of presentation in Moyamoya disease. Cortical blindness is very rare presenting symptom of Moyamoya disease. Progressive visual loss and homonymous anopsia has been described previously, but this case had acute visual loss.
**Key words**: Moyamoya disease, Cortical blindness, Angiography

## Introduction

Moyamoya disease is a rare progressive vasoocclusive disorder of an unknown etiology. It is characterized by progressive stenosis of terminal portions of internal carotid arteries bilaterally, and the main trunks of Anterior and Middle Cerebral Artery, and is associated with collateral vessels at the base of the brain ('Moyamoya' vessels). The term Moyamoya is a Japanese word which means "a puff of smoke". It was coined by Suzuki and Takaku in 1969 to describe the angiographic appearance of the collateral vessels at the base of brain in a group of 21 patients with internal carotid arterial occlusion. (16)

The histopathology of the affected arteries demonstrate a fibrocellular thickening of the intima, proliferated smooth muscle cells, prominently tortuous and often duplicates internal elastic lamina, with no inflammatory or atheromatous involvement. (18)

Moyamoya disease can present as progressive visual loss and homonymous anopsia which has been described previously. (14, 15)

## Case Report

A 12 year old girl admitted to our department had history of loss of vision in both eyes within 3 hours. The vision improved slightly after 10 days with perception of hand movements at 1 foot. Brain CT showed small multiple infarcts and large bilateral parieto-occipital lobe infarcts (Figure 1a) and brain T2W MRI showed flow voids around midbrain (1b).

Cerebral digital substraction angiography revealed occlusion of both supraclinoid internal carotid artery (Figures 2a and 2b) with collateral circulation from multiple enlarged lenticulostriate (Figure 2c) and thalamoperforating arteries (Figure 2d)

suggestive of Moyamoya disease. The detailed laboratory workup did not reveal any cause of progressive arteriopathy. Cortical blindness is very rare presenting symptom of Moyamoya disease. Moyamoya disease can present as progressive visual loss and homonymous anopsia which has been described previously, but this case had acute visual loss.

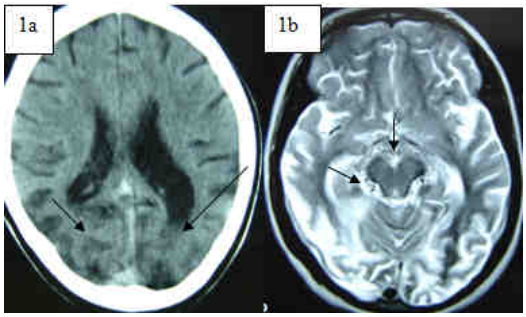


**Figure 1** - Brain CT shows bilateral parieto-occipital lobe infarcts (1a). Brain MRI shows flow voids around midbrain (1b)

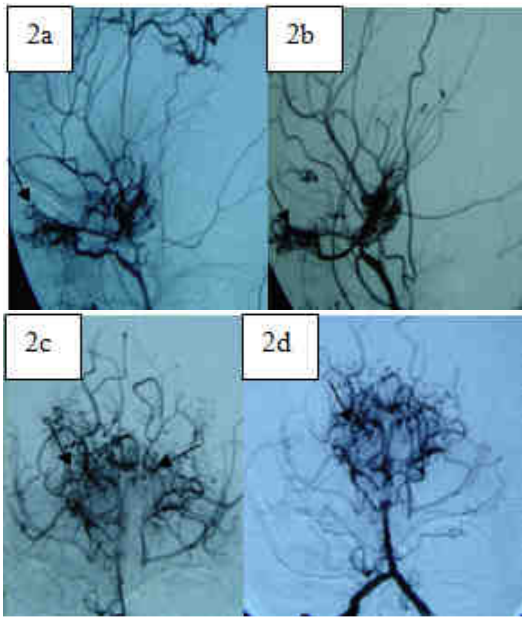


**Figure 2** - Cerebral digital substraction angiography revealed occlusion of both supraclinoid internal carotid artery (2a & 2b) with collateral circulation from multiple enlarged lenticulostriate (2c) and thalamoperforating arteries (2d) suggestive of Moyamoya disease

## Discussion

Moyamoya disease, first described by Takeuchi and Shimizu, is a rare cerebrovascular condition characterized by progressive stenosis of bilateral internal carotid arteries with compensatory formation of extensive collateral circulation by the dural, leptomeningeal and other perforating blood vessels giving rise to the typical "a puff of smoke" appearance in angiogram (17).

Moyamoya disease is reported to primarily present at an early age, typically less than 10 years, and is more common in females (M:F 1:1.8). However, a second peak in the fourth decade has also been described in the literature. (10) Etiology of Moyamoya disease is controversial. Even though several linkage studies have shown promising relations with gene loci (7, 8), no specific locus has yet been identified. However, these linkage studies, along with the familial occurrence of Moyamoya, point towards a probable genetic basis underlying its etiology. Investigations into understanding the pathogenesis of Moyamoya disease have shown involvement of the CSF basic Fibroblast growth factor with receptor up regulation, and TGF beta 1 in altering the cerebral vasculature (2, 4, 12). Intimal thickening has also been postulated resulting from altered permeability owing to enhanced prostaglandin release from the arterial smooth muscle. (19)

The clinical features of patients with

Moyamoya disease reflect the anatomic territory of the brain affected by the diseased vessel. Yamaguchi et al. in the Annual Report for the special working group of Welfare Ministry for Moyamoya in 1979 described four major types of Moyamoya disease according to clinical manifestations; the hemorrhagic type, the infarction type, TIA type, and epileptic type; with the first two types being the most common. (11)

Despite the reported risks associated with conventional angiography (5), it is still considered the gold standard for diagnosing Moyamoya disease. MR angiography proved to be a helpful diagnostic tool identifying sites of stenosis and demonstrating the collateral vessels at the base of the brain. As a noninvasive procedure, it has been described in the literature as a promising alternative to classical angiography for this arterial disease (16).

Irrespective of the radiological method used for diagnosis, conditions like von Recklinghausen's disease, Down's syndrome, autoimmune vasculitis, head trauma, meningitis and brain tumors may have a similar angiographic picture and hence can possibly confuse the diagnosis. Also, postpartum cerebral angiopathy and inflammatory angiopathy are two important differential diagnoses that may present with cerebral ischemia and imaging consistent with intracranial vasculopathy mimicking Moyamoya disease. (9, 13)

Four types of surgical procedures have been described indirect procedures including encephaloduroarteriomyosynangiosis, direct revascularization via the superficial temporal artery and the middle cerebral artery bypass, combined approaches and rarely, denervation of cerebral vasculature. In general, pediatric cases benefit from indirect revascularization procedures and the direct bypass is useful in most of adult cases. (6)

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