

# Moyamoya Disease in an 8-Year-Old Boy Presenting With Weakness

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**Abstract:** Moyamoya disease is a progressive, unilateral, or bilateral carotid artery stenosis of unknown etiology. It often presents in children as a transient ischemic attack, with a focal neurological deficit. This case describes an 8-year-old boy who presented with left-sided weakness secondary to moyamoya vasculopathy.

**Key Words:** moyamoya disease, transient ischemic attack, vasculopathy, magnetic resonance angiography

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## CASE

An 8-year-old white boy with a history of attention deficit hyperactivity disorder was transferred from an outpatient urgent care facility after a new onset of left-sided weakness. According to the patient and his parents, he had been having 2 to 3 days of upper respiratory tract infection symptoms such as cough, runny nose, and congestion as well as a low-grade fever. The morning of presentation, he awoke with a headache described as frontal in location and dizziness. He went to school and developed left upper extremity pain that over the course of the next several hours progressed to left upper and lower extremity weakness, which had resolved before his presentation to the urgent care. He denied photophobia, phonophobia, and vomiting.

The patient had a history of headaches, with no migrainous qualities, that he attributed to his Adderall. He did have a family history of migraine headaches and a stressor related to the recent separation of his parents.

On arrival, approximately 6 hours after the onset of symptoms, he was well appearing, in no acute distress, and he reported his weakness and headache completely resolved. His vital signs were as follows: temperature 37.6°C, heart rate 140 beats per minute, respiratory rate 28 breaths per minute, blood pressure 134/84, and weight 36.3 kg (90th percentile). His respiratory examination revealed some mild nasal congestion and clear lungs. Cardiovascular and abdominal examinations were normal, and he was hemodynamically stable. His neurological examination was normal including normal fundi and cranial nerves as well as strength 5/5 bilaterally and normal gait. He was alert and oriented to time, person, and place with a Glasgow coma scale score of 15.

A head computed tomography scan was obtained in the emergency department to investigate his transitory weakness and showed bilateral subcortical white matter foci in the frontal lobes with the left side more predominant (Fig. 1). This was interpreted by the radiologist as a demyelinating process, with a differential diagnosis of possible acute demyelinating enceph-

alomyelitis. He was admitted to the neurology service and magnetic resonance imaging (MRI) was obtained the following day showing moyamoya vasculopathy (Fig. 2). There was occlusion of the left carotid terminus and near occlusion of the right carotid terminus with associated lenticulostriate collateral vessels (Fig. 2A). There was also chronic left frontal white matter encephalomalacia and gliosis consistent with remote ischemic injury (Fig. 2B). There was no evidence of acute ischemic injury.

The patient had unremarkable complete blood cell count and liver function studies. Coagulation studies demonstrated a mildly prolonged partial thromboplastin time of 40.5 seconds (reference range, 25.9–35.6 seconds). Neurosurgery evaluated the patient and recommended a medrol dose pack and daily aspirin, and the patient was discharged home on those medications. He did not experience any further focal neurological changes during the admission. Outpatient neurosurgery follow-up was to occur within 1 month to discuss surgical options.

## DISCUSSION

A child that presents to the emergency department with an acute, focal neurologic deficit, especially one that has resolved, poses a diagnostic challenge. Although transient ischemic attacks (TIA) or stroke is 1 cause of a neurologic deficit, it is relatively rare with an incidence of 2.1 to 13.1 per 100,000 children per year.<sup>1</sup> Much more common are TIA or stroke “mimics” such as seizure or postictal deficit, tumor, infection, or complicated migraine. It has recently been shown that many disorders mimic childhood stroke including acute demyelinating encephalomyelitis, encephalitis, metabolic stroke, and postinfectious cerebellitis. History and clinical presentation have not been shown to be sufficient to distinguish benign disorders and serious problems.<sup>2</sup> Expedited MRI is a necessary tool for evaluating these patients and identifying disorders such as moyamoya vasculopathy.

## Epidemiology and Pathophysiology

Moyamoya disease was first reported in 1957 by Takeuchi and Shimizu as hypoplasia of 1 or both of the internal carotid arteries.<sup>3</sup> The Japanese term moyamoya is translated as a smoky, hazy, or vague state like cigarette smoke, which is used to describe the abnormal vascular networks seen.<sup>4</sup> This appearance arises from the vascular collateral networks that develop secondary to the bilateral stenosis or occlusion of the carotid arteries at the circle of Willis.

Overall incidence of moyamoya disease in the United States has been reported as 0.086 per 100,000, which is lower than the 0.35 per 100,000 reported in Japan.<sup>5</sup> Among ethnic groups within the United States, Asians have the highest incidence rates, 0.28 per 100,000. Although the disease is distributed among all age groups, onset predominates in those younger than 10 years (50%). The female-male ratio was 1.8:1.<sup>6</sup>

Although the cause of moyamoya disease is unknown, a high incidence in the Asian population and a familial occurrence of 10% makes a genetic etiology likely. Familial moyamoya disease has

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**FIGURE 1.** Computerized tomographic scan without contrast, axial view, showing left subcortical white matter foci (arrow).

been associated with a variety of chromosomes and is thought to have autosomal dominant transmission.<sup>7</sup> This syndrome has also been associated with a variety of medical conditions such as sickle cell disease, Neurofibromatosis Type 1, Down syndrome, Alagille syndrome, Williams syndrome, and posterior fossa brain malformations, facial hemangiomas, arterial/cardiac/eye abnormalities, sternal clefts, and supraumbilical abdominal raphe (PHACES) syndrome.<sup>8</sup> This association has led some to develop the term "moyamoya phenomenon" to distinguish from the idiopathic form.

### Clinical Presentation and Diagnosis

Moyamoya vasculopathy is a progressive, unilateral, or bilateral carotid artery stenosis, frequently with asymmetric onset and of unknown etiology. Presenting symptoms and signs are mainly focal neurological deficits (hemiparesis, monoparesis, and

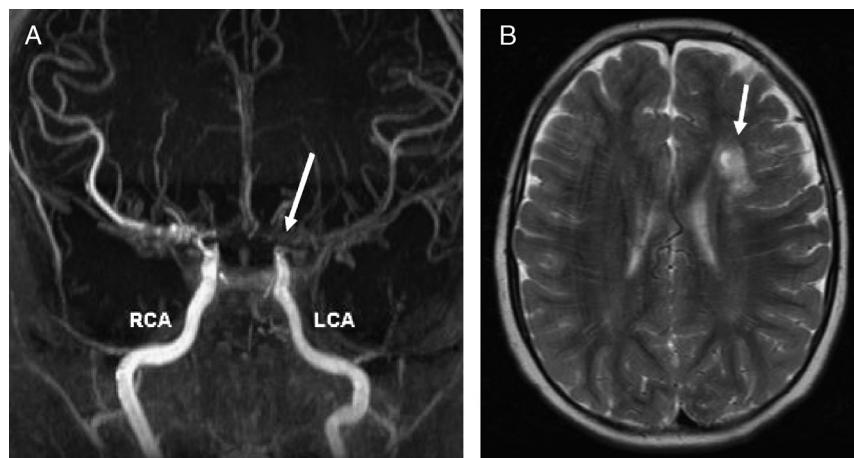
sensory disturbances) related to cerebral ischemia or hemorrhage. Ischemia is frequently manifested as TIAs and can occur repeatedly and can alternate sides because the vascular lesions are bilateral. Classically, these alternating and repeated TIAs, particularly during exercise, are the first manifestation of the disease.<sup>9</sup> More nonspecific neurologic symptoms can also be manifested such as headaches, seizures, or progressive decline in cognitive function.<sup>9</sup> As the illness progresses, there can be cerebral infarction resulting in permanent neurologic deficits. Intracerebral hemorrhage has been reported as rarely occurring in children and is a much more common presentation in adults.<sup>4</sup>

The diagnosis of moyamoya is made by visualizing the characteristic angiographic appearance of the carotid vessels. Cerebral angiography is the gold standard for diagnosis, and this is most commonly done with magnetic resonance angiography (MRA). The characteristic findings include stenosis or occlusion at the region of the distal internal carotid arteries and the origin of the middle and anterior cerebral arteries. However, there can be a wide variety of cerebral vessel involvement, including arteries of the posterior cerebral circulation.<sup>10</sup> In addition, there are frequently abnormal vascular networks as well which function as collateral pathways. Bilateral involvement has been referred to as a definite case and unilateral involvement a probable case.<sup>4</sup> Computed tomography is usually not helpful for the acute phase of the disease. Magnetic resonance imaging is superior for detecting small ischemic lesions.

### Treatment and Prognosis

Acute early treatment of symptoms of stroke or known moyamoya disease should follow the American Heart Association guidelines for all types of ischemic stroke.<sup>11</sup> Management techniques include airway support and ventilatory assistance as needed, keeping the head of the bed flat, supplemental oxygen, antipyretics to maintain normothermia, permissive hypertension to maintain adequate cerebral perfusion pressure, normal saline to correct hypovolemia, and maintaining normoglycemia.

Several definitive treatments have been considered for moyamoya disease and are divided into medical and surgical approaches. Medical treatments on the whole, such as vasodilators, steroids, and anticoagulants, have been shown to be ineffective.<sup>12</sup> If there is an underlying cause for moyamoya, such as sickle cell disease, it is important to treat that disease. Pediatric moyamoya disease is mainly treated with a variety of



**FIGURE 2.** (A) MRA showing occlusion of the left carotid artery terminus and near occlusion of the right carotid artery terminus. The arrow shows the point of occlusion in the left carotid artery. (B) T2 MRI showing left sided white matter signal change associated with an area of old ischemia (arrow).

surgical approaches designed to revascularize ischemic cerebral cortex. The first group is categorized as direct procedures such as direct anastomosis of arteries between extracranial and intracranial circulations. There are also indirect procedures, such as encephaloduroarteriosynangiosis and pial synangiosis, the goal of which are to encourage collateral vessel formation by directly placing dura, temporalis muscle, or omentum onto the brain surface.<sup>13</sup> It has been found that most pediatric patients have symptomatic benefit as a result of pial synangiosis with stroke rates of 3.2% in the 1 year postoperative period.<sup>14</sup>

Long-term outcomes of pediatric moyamoya disease have varied among reports, likely due to differences in disease types, age of onset, and follow-up periods but overall, long-term outcomes are generally poor. Risk factors indicating poor prognosis have included early onset age, non-TIA presenting symptoms, and hypertension.<sup>15</sup>

## CONCLUSIONS

Cerebrovascular disease, such as moyamoya, is uncommon in children but can result in significant morbidity and mortality. Emergency department physicians are often the first to evaluate these children. It is important to recognize the common presenting features, which have been found to differ based on age.<sup>16</sup> In addition, there is often a delay in diagnosis that can increase the chances of a poor outcome.<sup>17</sup> Given that most stroke syndromes in children are related to congenital or acquired nonatherosclerotic cerebrovascular disease, MRA is always a useful part of the initial imaging workup.<sup>18</sup>

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