A Rare Case of Moyamoya Disease in a 20-year-old Puerto Rican Female U.S. Soldier

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Moyamoya disease is a progressive, occlusive pathology involving the cerebral vasculature with particular involvement of the circle of Willis and its tributaries. The cause of moyamoya disease is unknown, but is believed to be hereditary. Females 20 to 39 years old with moyamoya represent 0.5% of all acute cerebral ischemia and infarcts with risk factors including smoking, estrogen-containing birth control use, coagulopathy, neoplasm, and congenital malformation. This case reports on a 20-year-old Puerto Rican female U.S. soldier with a 1-year history of migraine headaches with worsening right retro-orbital pain, blurred vision, and photophobia. The patient had minimal unilateral neurological deficits despite evidence of significant cerebral infarction on non-contrast computed tomography. Other neuroimaging findings were consistent with moyamoya disease with confirmation via cerebral angiography. This case details the process of diagnosis and treatment as well as discussing its incidence, identification, and treatment options. [*P R Health Sci J 2014;33:197-199*]

Key words: Moyamoya, Cerebrovascular occlusive disease, Ischemia

Moyamoya disease is a rare, progressive cerebrovascular disorder secondary to arterial obstruction at the proximal Circle of Willis arteries. More common in pediatric patients, the first symptom is often stroke or recurrent transient ischemic attacks frequently accompanied by muscular weakness, seizures, or unilateral paralysis. Adults, however, frequently experience a nontraumatic intracerebral hemorrhage theorized to be secondary to formation of extensive collateral vessels with an increased proclivity of rupture. Most individuals with this disorder have disturbed consciousness, speech deficits, sensory and cognitive impairments, involuntary movements, and vision problems (1, 2).

Case Report

A 20-year-old female soldier of Puerto Rican ancestry presented to the Womack Army Medical Center, Fort Bragg, NC emergency department (ED) following 7 days of right retro-orbital pain, 2 days of blurred vision and photophobia, as well as 1 day of nausea and neck pain. Her headaches increased during the previous month and intensified 4 days prior to presentation. At evaluation, mild paresthesia in her left arm was noted.

Her medical history revealed 1 year of migraine headaches lasting from 2 to 72 hours with unilateral throbbing pain and occasional blurred vision, generally controlled with Excedrin®. She had started estrogen-containing oral contraceptive medication 6 months prior without adverse effects.

The patient's Puerto Rican mother died of myocardial infarction at age 37 and her father, an African American, has a history of diabetes mellitus, hypertension, and dyslipidemia; there was no family history of coagulopathies. Her 3 siblings, including an identical twin sister, are all in good health. The patient worked as a water purification and fueling specialist and had not deployed overseas nor had any chemical exposures during her 18 months in the Army.

Her physical exam was unremarkable. A non-contrast head computed tomography (CT) revealed right subacute middle cerebral artery region ischemia without signs of hemorrhage. Other neuroimaging findings – magnetic resonance imaging (MRI), magnetic resonance angiogram (MRA), magnetic resonance venography (MRV), and computed tomography angiography (CTA)– were consistent with moyamoya disease and confirmed by cerebral angiography (Figures 1, 2).

The patient was transferred to the University of North Carolina neurosurgical department where her neurological findings resolved without intervention. She was medically discharged from the Army and underwent extracranial-intracranial bypass surgery to reduce the risk of stroke recurrence by improving the flow of blood to the blocked artery with revascularization. The patient has had no relapse of symptoms or persistent neurological deficits.

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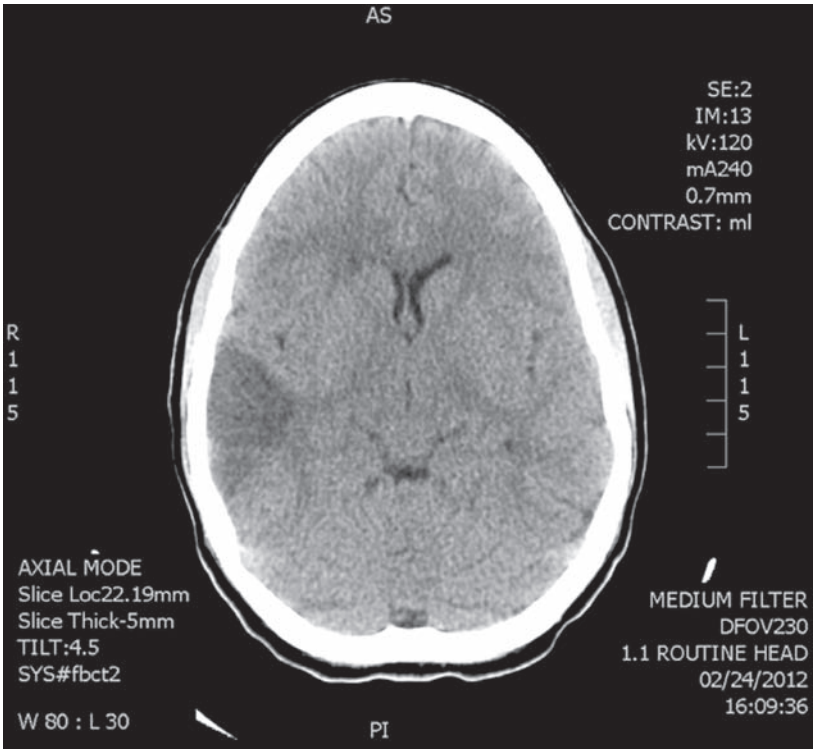

Figure 1. Computed tomography non-contrast of head revealing late acute to subacute middle cerebral artery cerebrovascular accident

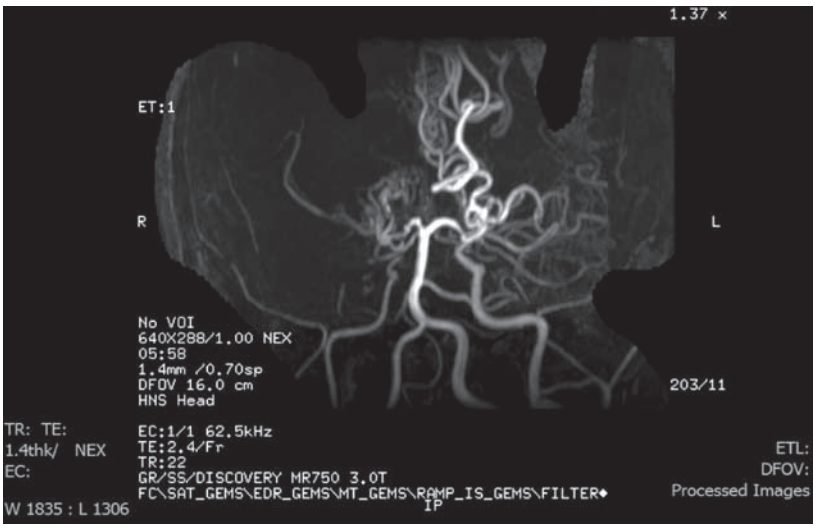

Figure 2. Magnetic Resonance Angiography (MRA) image

Discussion

The prevalence and incidence of moyamoya disease varies between ethnic populations, including Caucasians, Asians, African Americans, Haitians, and Hispanics. While many cases have been reported in Japan (3), the disease has also been found in the United States (4-6) and Europe (7). The disease has a female-to-male ratio of 1.8:1. Age of moyamoya onset ranges from 6 months to 67 years with the highest peak in the first decade.

The first case of moyamoya disease was published in 1957 (8). The Japanese term "moyamoya" was later coined to describe the hazy, cloudy "puff of smoke" appearance of the network of dilated, abnormal microvasculature occurring in the region of the circle of Willis (2). The stages of angiographic progression were outlined as knowledge of the disease increased, from stage 1– narrowing of the carotid artery – to stage 6, when moyamoya vessels disappear and the external carotid arteries supply collateral flow. Although presumed to be hereditary, a clear etiology of moyamoya disease is unknown. Family history has been observed in 10% of moyamoya patients (9) and familial moyamoya disease may be autosomal dominant with incomplete penetrance that depends on age and genomic imprinting factors (10). Genetically, susceptibility loci have been found (11).

The outcome of moyamoya disease depends on the severity and nature of the hemorrhage; prognosis depends on recurrent ischemic attacks. Death usually follows intracranial hemorrhage with mortality rates about 10% in adults and 4.3% in children. Fifty to 60% of affected individuals experience a gradual deterioration of cognitive function, presumably from recurrent cerebral ischemia and infarction. Patients who present for treatment while symptoms are evolving have a better prognosis compared to those who present with static symptoms, usually indicative of a cerebral infarction and total loss of functional tissue.

Misdiagnosis and delayed diagnosis of moyamoya disease are common. Stroke is often the result of progressive or abrupt atherosclerotic occlusion of the carotid arteries which occurs in older adults who have several risk factors. In moyamoya disease, however, the only risk factor may be genetic. The diagnosis of stroke may be delayed as alternative etiologies are explored, such as hypercoagulable states.

Currently, there are few pharmacologic therapies for moyamoya disease. If nontraumatic intracerebral hemorrhage has occurred, then management of hypertension (if present) is imperative. In cases of severe cerebral ischemia and infarction,

intensive care unit monitoring is indicated until the patient's condition stabilizes. Anticoagulation or antiplatelet agents should be considered after ischemic attack. Inpatient medical recommendations include: aspirin use (2-5mg/kg in children or 50-100mg in adults) but no long-term anticoagulation; avoidance of hypocarbia associated with hyperventilation, hypotension, hypovolemia, and hyperthermia; isotonic fluid administration at 1.25-1.5 times maintenance rate; and oxygen supplementation (3).

Several factors contributed to the successful management of this patient. CT imaging was obtained early due to the patient's evolving migraine symptoms. Imaging studies were heavily utilized in diagnosis. MRI results reported "dilated collateral vessels in the basal ganglia and thalamus can be demonstrated as multiple punctuate flow voids, a finding which is virtually diagnostic for moyamoya." Although the criteria (Table 1) for the diagnosis of moyamoya are based on both angiography and magnetic resonance (12), recognition of this disease is often limited to those who consider moyamoya in the differential for patients with hemiparesis, monoparesis, sensory impairment, headaches, dizziness, seizures, or numerous other central neurological abnormalities.

Table 1. Criteria for the diagnosis of moyamoya disease in the absence of other underlying etiologies*

Angiography	Magnetic Resonance Angiography
Stenosis or occlusion at the terminal internal carotid artery and/or proximal anterior cerebral artery or middle cerebral artery	Stenosis or occlusion at the terminal internal carotid artery and/or proximal anterior cerebral artery or middle cerebral artery on magnetic resonance angiography
Abnormal vascular network in the vicinity of the lesion in the arterial phase	Abnormal vascular network in the basal ganglia on magnetic resonance angiography
Both seen BILATERALLY	Both seen BILATERALLY

*From: Fukui M, et al. Guidelines for the diagnosis and treatment of spontaneous occlusion of the circle of Willis ('Moyamoya' disease). Clin Neurol Neurosurg 1997;99:S238-S240.

Our patient was quickly diagnosed with moyamoya disease and treated. This was attributed in part to current diagnostic tests in place. Further research is necessary to recognize the symptoms and risk factors that suggest moyamoya. Once identified, additional efficacious treatment options can be sought.

Resumen

La enfermedad de moyamoya es una patología progresiva y oclusiva que involucra el sistema vascular cerebral; involucra particularmente el círculo de Willis y sus tributarios. Se desconoce la causa de la moyamoya, pero se cree que es hereditaria. Las mujeres de 20 a 39 años de edad con moyamoya representan el 0.5% de todos los infartos e isquemias cerebrovasculares agudos con factores de riesgos que incluyen fumar, uso de anticonceptivos que contienen estrógeno, coagulopatía, neoplasma, y malformación congénita. Este caso reporta una puertorriqueña, soldado de E.U., que padece hace un año de migrañas con peor dolor retro orbital de la derecha, visión borrosa, y fotofobia. La paciente tuvo déficits neurológicas mínimas a pesar de la evidencia de infarto cerebral en tomografía computarizada sin contraste. Otros hallazgos de neuroimágnes eran consistentes con la enfermedad de moyamoya confirmado vía angiografía cerebral. Este caso detalla el proceso de diagnóstico y tratamiento y discute su incidencia, identificación y opciones de tratamientos.

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