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A case with unusual stroke and fulminant outcome in a Hispanic male

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Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
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Patient: Male, 42
Final Diagnosis: Moyamoya disease (MMD)
Symptoms: Aphasia • concentration difficulty • dysarthria • personality change
Medication: —
Clinical Procedure: —
Specialty: Radiology

Objective: Rare disease

Background: Moyamoya disease (MMD) was first described in 1957 as "hypoplasia of the bilateral internal carotid arteries." The characteristic appearance of the associated network of abnormally dilated collateral vessels on angiography was later likened to "something hazy, like a puff of cigarette smoke," which, in Japanese, is *Moyamoya*. This paper describes the fulminant course of the disease in a Hispanic male involving the corpus callosum.

Case Report: A 42-year-old Hispanic male with progressive aphasia, slow mentation, and sudden onset of sensorimotor symptoms with gait disturbance was found to have multiple intracranial supratentorial infarcts of variable stages of evolution involving, but not limited to, the anterior corpus callosum, followed by rapid development of further infarcts. Angiography demonstrated right ACA occlusion, left supraclinoid ICA occlusion with a Moyamoya pattern of collateralization, and diffuse arteriopathy. A fulminant course ensued and the patient did not survive the acute phase of ischemic disease.

Conclusions: Moyamoya disease may rarely present in North American Hispanic males, with advanced atypical clinical and imaging features involving the anterior corpus callosum and having a fulminant course.

Key words: infarction • corpus callosum • moya moya disease

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Background

Moyamoya disease is an uncommon, progressively occlusive intracranial arteriopathy of the anterior cerebral circulation, typically presenting as an acute infarct with a high risk for subsequent strokes [1]. Although the typical phenotype affects Japanese children, North Americans affected by the disease are typically adults of Asian American, Black, or Hispanic ethnicity [2].

The cause of Moyamoya disease is unknown. The high incidence among Asian populations, together with a familial occurrence of approximately 105 of cases, strongly suggests a genetic cause [2].

Accumulating evidence suggests that the RNF213 gene on chromosome 17q25.3 is an important susceptibility factor for MMD in East Asian populations [3–7]. In a report from Japan, the c.14576 G>A variant of RNF213 was found in 95% of 41 patients with familial MMD, 79% of 163 patients with sporadic MMD, and 2% of 283 normal control subjects [5]. Other studies have linked familial Moyamoya disease to chromosomes 3p24.2-p26, 6q25, 8q23, and 12p12 [8–10].

The vascular pathology usually worsens, with extensive intracranial large artery occlusion and collateral circulation. Patients often suffer cognitive and neurologic decline due to repeated ischemic stroke or hemorrhage [3].

Clinical features can be insidious and presentation may be delayed, especially in this population of North Americans. Given the progressive nature of the disease and diffuse involvement, late presentation of Moyamoya disease may present with an atypical clinical and imaging pattern of vascular disease.

The natural history of Moyamoya may be different in North America, where patients tend to present with later onset and ARE less likely to have hemorrhagic stroke [9–12].

However, regardless of the course, Moyamoya inevitably progresses in the majority of patients [13]. A 2005 report indicated that the rate of disease progression is high, even among asymptomatic patients, and that medical therapy alone does not halt disease progression [14]. It has been estimated that up to two-thirds of patients with Moyamoya have symptomatic progression over a 5-year period; the outcome is poor without treatment [15–17]. In contrast, the estimated rate of symptomatic progression is only 2.6% after surgery, according to a meta-analysis involving 1156 patients [18].

We describe the first case, to our knowledge, of advanced Moyamoya disease in a Hispanic male presenting with an acute infarction of the rostrum and body of the corpus callosum with a fulminant progression.

Case Report

A 42-year-old, right-handed Hispanic male without significant past medical history presented with a 4-week history of slow mentation, difficulty concentrating, and change in personality. He also complained of bilateral lower extremity weakness causing difficulty walking, as well as a presyncopal episode and sudden onset of paraesthesias and numbness involving his right body side 3 days before admission. Other symptoms included intermittent nausea and vomiting.

Findings upon examination included slow mentation, decreased concentration, weakness of the right lower half of the face, dysarthria, and mild weakness in the lower extremities without sensory level or sphincter dysfunction. The patient had full strength in his upper extremities, diminished reflexes throughout, and intact sensation. No cerebellar dysfunction, nystagmus, or Babinski signs were present. Laboratory tests for systemic autoimmune, inflammatory, and infectious diseases, and neoplasm were all negative.

Initial unenhanced head CT demonstrated hypodense lesions involving the body and genu of the corpus callosum and right basal ganglia. A subsequent brain MRI showed multiple supratentorial lesions predominately involving the rostrum of the corpus callosum, genu of the right internal capsule and basal ganglia and subcortical cingulate white matter, with variable degrees of enhancement associated with mass effect and restricted diffusion (Figure 1).

Considering the clinical and the radiological features, initial differential diagnoses included an acute demyelinating process *versus* primary CNS lymphoma, glioma, or acute/subacute ischemic infarct.

Cerebrospinal fluid analysis demonstrated slightly increased total protein and elevated CSF levels of myelin basic protein, without evidence for oligoclonal bands. High doses of intravenous methylprednisolone (1 gm/day) were initiated; 72 hours after admission, the patient developed decreased level of consciousness, global aphasia, left gaze deviation, and dense right hemiplegia.

Cerebral angiography (Figure 2) demonstrated bilateral intracranial arteriopathy with occlusion of the supraclinoid segment of the left intracranial internal carotid artery distal to the takeoff of the left anterior choroidal artery, occlusion of the right anterior cerebral artery, and severe disease in the proximal right middle cerebral artery. The distal branches of the right middle and anterior cerebral arteries were mainly fed by collateral anastomoses from the anterior and posterior choroidal arteries, producing arterial

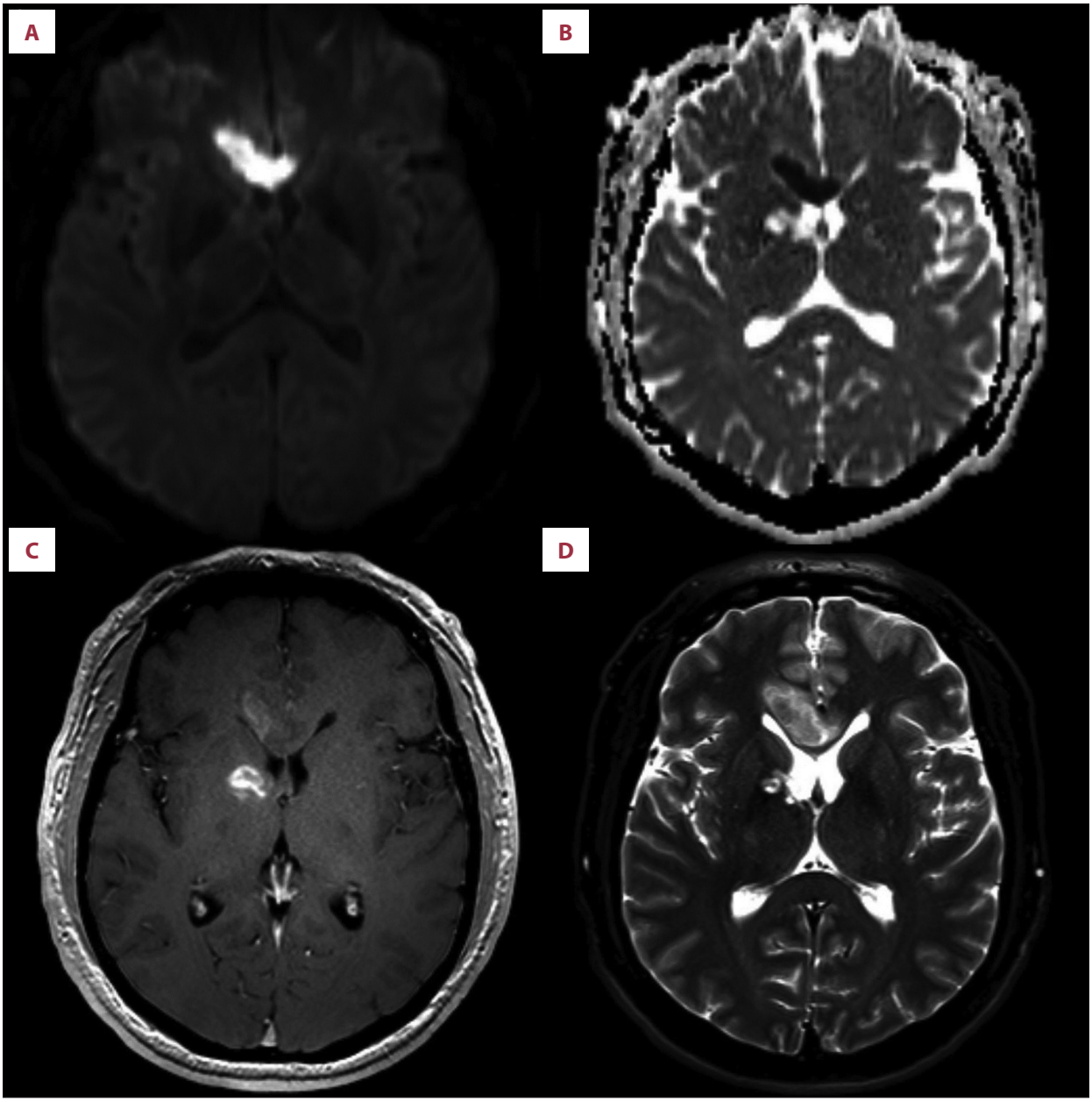

Figure 1. Brain MRI. (**A, B**) diffusion weighted sequence and apparent diffusion coefficient map, respectively, demonstrate a focal area of restriction of diffusion involving the right rostrum, genu and body of the Corpus Callosum with mild enhancement on post-gadolinium T1W sequence (**C**) and moderate edema and local mass effect on T2W sequence (**D**). A second lesion involving the right basal ganglia and genu of the right internal capsule demonstrates incomplete peripheral enhancement with central necrosis, without mass effect or restriction of diffusion.

blush ("puff of smoke") consistent with an angiographic Moyamoya pattern.

Subsequently, he developed a massive infarction involving almost the entire left cerebral hemisphere plus the bilateral anterior cerebral arterial territories, with associated cerebral edema, midline shift complicated by brain herniation, and eventually death 11 days after admission.

Discussion

Moyamoya disease is an idiopathic progressive intracranial occlusive arteriopathy that typically involves the anterior cerebral circulation. Pathologic examination of the intracranial arteries failed to prove atherosclerotic, inflammatory, or degenerative (amyloid) changes. The cause of the occlusion in Moyamoya disease remains idiopathic and may be multifactorial [1].

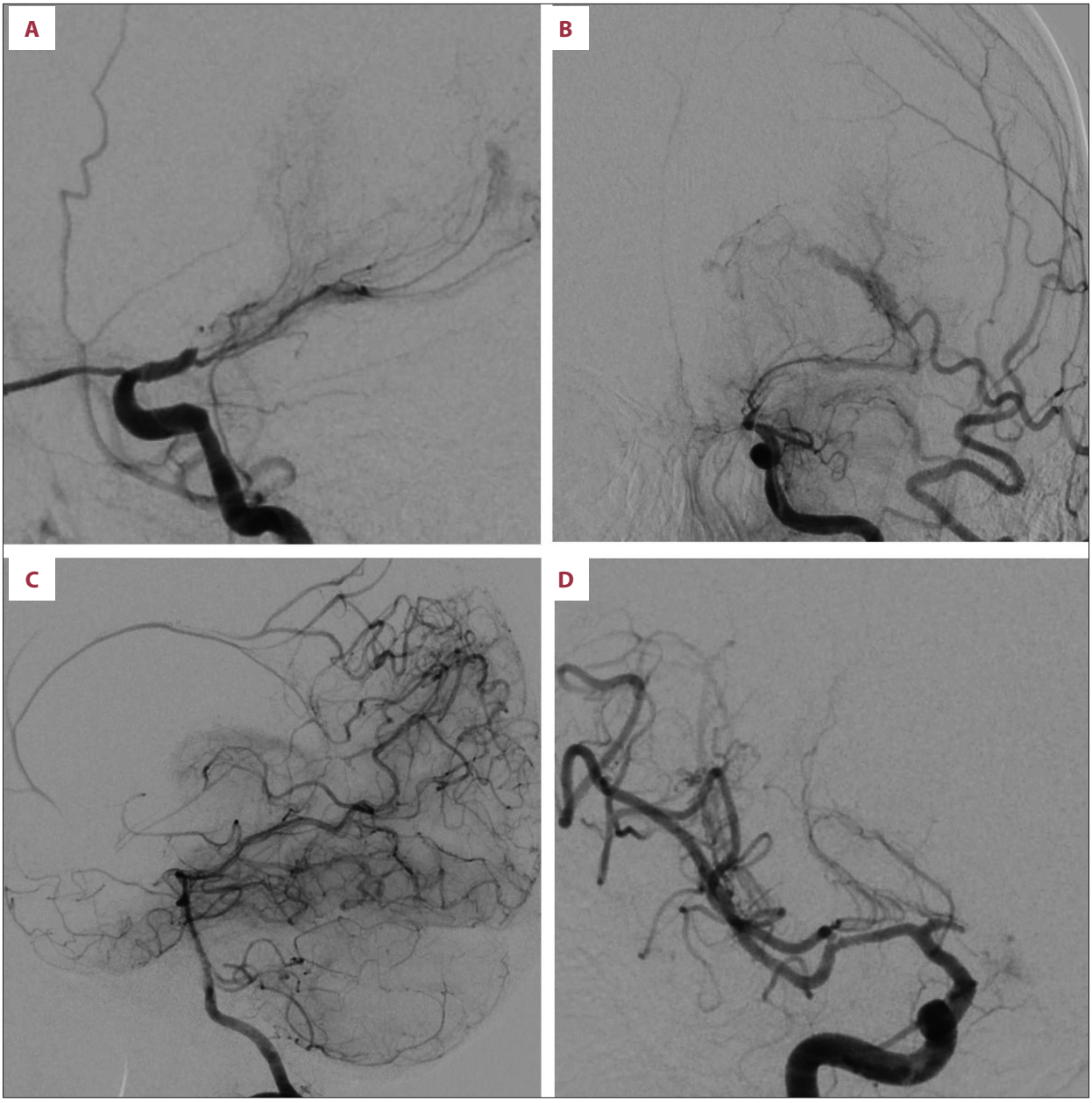

Figure 2. Cerebral angiography. Biplanar cerebral angiography of the left internal carotid artery ((**A, B**) lateral and AP respectively) demonstrated complete occlusion of the left supraclinoid internal carotid artery with a patent left anterior choroidal artery supplying lenticulostriatal vessels giving the angiographic appearance of a "puff of smoke," suggestive of Moyamoya angiographic pattern. Collateral flow from the right posterior choroidal artery supplying the pericollosal, frontopolar and callosmarginal arteries demonstrated on lateral view on selective angiogram of the left vertebral artery (**C**). Intracranial view of the right internal carotid artery in the AP projection (**D**) demonstrates an irregular M1 segment and complete occlusion of the mid A1 segment of anterior cerebral artery. Cortical branches also appeared irregularity consistent with a proliferative intracranial vasculopathy.

Associations with *loci* on chromosomes 3, 6, 8, and 17, as well as specific HLA haplotypes, have also been described [3–7,19].

The disease can be complicated with ischemic and hemorrhagic cerebrovascular events. The hallmark feature of Moyamoya is a distinct angiographic pattern of collateralization producing a pathognomonic "puff of smoke" (Moyamoya). Certain diseases can produce a similar angiographic pattern. The Moyamoya syndrome can be associated with severe intracranial atherosclerotic disease, sickle cell disease, neurofibromatosis type I, cranial radiation therapy, and trisomy 21 [1,20].

As first described, the Asian phenotype of Moyamoya disease is typically a disease of childhood, whereas the North American form can present in adults in their mid-40s [2]. Reported sex predominance favors females approximately 2:1 [20]. Overall incidence of Moyamoya is 0.086 cases per 100 000 persons. Moyamoya is the most common pediatric cause of cerebrovascular disease in Japan, with a prevalence of 6.03 per 100 000, while in the USA it is thought to be lower [21]. The incidence among all patients with Moyamoya in Europe appears to be about 1/10th of that observed in Japan [22].

An increase in the prevalence of Moyamoya has been demonstrated among non-Asian populations. The reported occurrence in the Caucasian population in the US is the highest compared to other populations, but Caucasians represent 72.4% of the US population. In general, Moyamoya disease is observed among the various races based on their proportions of the total US population [23,24].

Familial occurrence of the disease in the US is approximately 2%, but in Japan a family history of Moyamoya disease is present in 12.1% of patients. Environmental factors can be considered as contributing factors to this observation [21,23].

Overall, amongst the North Americans affected by Moyamoya, Asian Americans have the highest incidence, followed by blacks and Hispanics. There is a presumed higher incidence in non-white and blacks compared with whites [2,20].

Moyamoya disease is a chronic and progressive intracranial arterial disease that frequently affects both hemispheres, leading to infarcts or TIAs and intracerebral hemorrhages. Given the rich vascular supply of the corpus callosum, infarcts involving the anterior corpus callosum are rare and often present a diagnostic challenge for clinicians and radiologists alike [25,26]. The vascular supply of the corpus callosum is derived from 3 primary sources forming a rich anastomotic network, making infarcts in this region uncommon. The anterior communicating artery usually gives rise to the subcallosal and median callosal arteries, which supply the anterior portions of the corpus callosum. The pericallosal artery, a continuation of the anterior cerebral artery, typically gives rise to the recurrent cingulocallosal artery from which the callosomarginal artery arises, supplying the body of the corpus callosum. The posterior pericallosal artery, typically arising from the posterior cerebral artery, supplies the splenium of the corpus callosum [27]. On imaging, infarcts of the corpus callosum often demonstrate a significant mass effect and a variable degree of enhancement [28], mimicking neoplastic or acute demyelinating processes. As in our patient who presented with a history of progressive aphasia, subsequent right-sided sensorimotor deficits and gait disturbance, sequelae of infarcts of the corpus callosum may be atypical and non-localizing, with symptoms frequently attributed to interhemispheric disconnection. Crossed aphasia has been described in right-handed patients with an infarct of the right anterior corpus callosum, attributed to crossed diaschisis [12]. Additionally dyspraxia, alien hand syndrome, and isolated gait disorder have specifically been ascribed to lesions in the anterior corpus callosum [28,29,32].

Conclusions

It is unusual for Moyamoya disease to present as acute infarct of the corpus callosum with a rapid progression and fulminant course resulting in multiple bilateral large lobar infarcts precipitating herniation and death within days [29]. The prevalence of Moyamoya disease appears to be increasing worldwide. To our knowledge, this clinical presentation of Moyamoya disease has not previously been described in the literature. Inclusion of occlusive intracranial arteriopathy as a differential diagnosis for lesions of the corpus callosum may allow for more rapid diagnosis [30]. Direct surgical revascularization procedures such as superficial temporal artery to middle cerebral artery bypass [STA-MCA] or indirect bypass surgery procedures that introduce external carotid flow into the internal carotid system can be used in certain cases [31]. While definitive treatment in the acute fulminant phase may be futile and not indicated, ascertaining the correct diagnosis may help provide a more accurate prognosis [33].

Disclosures

All participating authors in this study declare no financial, professional, or personal conflicts.

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