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Moyamoya disease: review and demographic description of a series of cases in Bogotá, Colombia

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Abstract

Introduction: *Moyamoya disease (EM) is an unusual pathology that presents with unilateral or bilateral occlusion of the supraclinoid portion of the internal carotid arteries accompanied by vascular neoformation as a compensatory method of arterial blood flow to the brain parenchyma.* **Objective:** *The objective of the study is to describe the experience in the diagnosis, management, and treatment of EM in a fourth level hospital in Bogotá, Colombia, in relation to the literature available in the databases.* **Methods:** *A retrospective study was carried out through the search in medical records, and collecting patients diagnosed with EM, demographic variables, risk factors, diseases associated with the diagnosis, treatment, and follow-up of the patients included in this study were obtained.* **Results:** *Six cases of different ages were collected, on average 24 years of age, a case with neurofibromatosis type 1 was found, and a case with a first-degree family history, the most frequent clinical presentations were headache, convulsion, and hemorrhagic or ischemic accidents. Arteriography was the gold-standard diagnosis; 83% of the patients received surgical treatment; no relapses or associated mortality was documented at follow-up; however, in 50% of the cases, some type of neurological deficit was identified.* **Conclusions:** *The clinical manifestations presented in our cases are compatible with those described in the literature, revascularization surgery was effective taking into account the sequelae and neurological status of the patients, we consider that a long-term follow-up of all cases is necessary to establish sequelae and recurrences.*

Keywords: *Ischemic stroke. Hemorrhagic stroke. Moyamoya disease. Moyamoya syndrome. Revascularization.*

Introduction

Moyamoya disease (MS) is a rare cerebrovascular condition in which there is a progressive occlusion of the terminal portion of the uni- or bilateral internal carotid artery and with this the formation of an abnormal cerebral vascular network[1]. First described in 1957 as a bilateral hypoplasia of the internal carotid arteries, it was observed in angiographic studies that the formation of collateral vascular networks resembled a distribution in cigarette smoke, which in Japanese translates to moyamoya (moyamoya sign)[1,2] unlike moyamoya syndrome (MS) which must be associated with a medical condition. Among the main ones are neurofibromatosis type 1, Down syndrome, sickle cell disease, and radiotherapy of the head or neck; on the other hand, those patients who do not have associated risk factors are said to have MS[3,4].

This pathology is more common in populations of Asian descent, with a prevalence in children of approximately 3/100,000 specifically reported in Japan[4]; however, an increasing trend in its incidence has been

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observed in America, studies in the United States suggest that the incidence is around 1/100,000 people, as well as an incidence in Europe in one tenth of Japan. MS has two peaks, the first around 5 years of age and the second at 40 years of age[1,5] and is more common in women than in men[6,7]. 15% of MS patients have a family history, identifying that there are genetic factors in relation to this pathogenesis where genome studies have assigned their locus of susceptibility to the RNF213 gene on chromosome 17q25.3 mainly in Japanese families[8].

Typically, MS is shown bilaterally, unilateral involvement also occurs, looking for risk factors for the contralateral progression of unilateral MS; in a multicenter cohort study conducted in Japan, it was concluded that both genetic (RNF213 gene mutation) and non-genetic (environmental) factors are associated with the contralateral progression of unilateral MS[3].

Its clinical presentation varies depending on age; however, in both adults and children, the signs and symptoms could be categorized into two groups, the main one being the ischemic event (transient or definitive)[5,9] and seizures followed by intracranial hemorrhage, more common in adults but also described in children in a lower percentage[2]; all of these are associated with headache, especially in children due to dilation of the meningeal and leptomeningeal collateral vessels[1,2].

The gold-standard (GS) diagnosis is cerebral arteriography with the image in "cigarette smoke" given by the collateral arterial formations, allowing its classification according to the criteria of Suzuki and Takaku[10,11] (Table 1), there are also complementary studies, which although they are not classified as GS, are of great diagnostic help, these being scans or brain resonance[4,7].

The mainstay of treatment is direct or indirect cerebral revascularization[1]; however, the use of anticoagulants is necessary to prevent the formation of emboli as a result of thrombi formed at the level of arterial stenosis[1,7].

Methods

A retrospective study was carried out where data were collected from patients diagnosed with MS or MS from medical records with the ICD-10 code I675. Demographic factors, risk factors, concomitant diseases, symptoms on admission, images performed, arteriography results, surgical and medical management, and finally early neurological sequelae were taken into account to perform a descriptive multivariate analysis (Table 2).

Table 1. Suzuki rating system

Grade	Definition
I	Terminal internal carotid artery stenosis
II	Home collateral vessels
III	Progressive carotid artery stenosis with progressive intensification of collaterals
IV	Development of collaterals from the external carotid artery
V	Intensification of collaterals from the external carotid artery with reduction of the moyamoya vessels
VI	Total occlusion of the internal carotid artery and disappearance of the moyamoya vessels

Results

Six cases were collected, with an average age of 24 years, and an equal distribution by female and male sexes (1:1) was found. Among the associated medical conditions for MS, a case with neurofibromatosis type 1 was described and in addition, another case with a maternal history of MS that, although it does not qualify as a medical condition, is relevant taking into account the aforementioned genetic studies. Cardiovascular risk factors were taken into account, including hypertension in 50% of cases, diabetes mellitus in 16%, which could have a slight impact as an environmental factor, the rest of the cases were classified as MS.

The predominant clinical picture at admission was headache with 33% of the patients, 33% due to epileptic seizures, 17% due to drowsiness, and 17% with loss of consciousness. The initial diagnostic studies were 50% simple brain tomography, 33% brain magnetic resonance imaging, and 17% brain angiography; From these images, four strokes were documented (66%) of which two patients had ischemic presentation and two hemorrhagic presentations, the rest of the patients did not present alterations in the diagnostic images of admission.

For the definitive diagnosis, cerebral arteriography was performed as BG in all cases, among which the bilateral presentation of moyamoya was most frequently observed in 83% and unilateral presentation in 17%, the presentation of stenosis in the posterior circulation (basilar artery and posterior cerebral artery) in two of the cases is striking. These are not commonly described findings in the literature. In the arteriography studies, the

Table 2. Study variables

Variable	No.	%
No. cases	6	
Media age (years)	24	
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Sex feminine	3	50
Masculine	3	50
Riesgo cardiovascular		
Hypertension arterial	3	50
Diabetes mellitus	1	17
Smoking	0	0
Dyslipidemias	0	0
Symptoms on admission		
Deficit	1	17
Headache	2	33
Seizures	2	33
Sleepiness	1	17
Loss of consciousness	1	17
Entrance image		
CT scan of the skull	3	50
MM brain	1	17
AngioMM brain	2	33
Image findings		
Ischemia	2	33
Hemorrhagic	2	33
No stroke	2	33
Parangiography	6	100
Interality Moya		
Unilateral	5	83
Bilateral	1	17
Susuki Rating		
I	0	0
II	0	0
III	1	17
IV	4	66
V	1	17
Treatment		
Medical	1	17
Surgical	5	83
Revascularization surgery		
Direct	1	17
Indirect	4	66

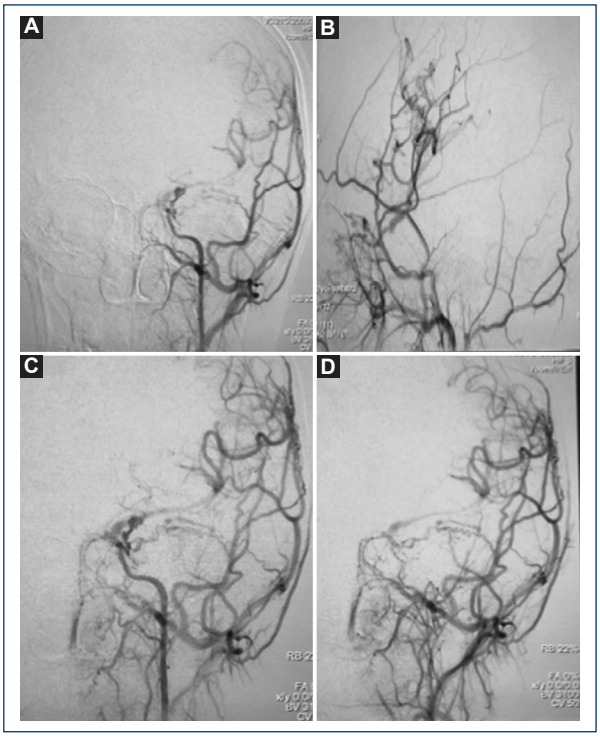

Figure 1. A and **C:** pre-operative subtraction angiography with supraclinoid stenosis of the internal carotid artery. **B** and **D:** post-operative control angiography, evidencing neovascularization after the performance of left arteriodurmyosinangiosis.

cases were cataloged according to the Suzuki and Takaku classification (11), with 67% being the Suzuki IV, 17% Suzuki III, and the remaining 17% Suzuki V.

In the management of the pathology, one patient received medical management with enoxaparin and five patients (83%) required revascularization; in four cases (67%), indirect revascularization surgery was performed with encephaloduroarteriosynangiosis and encephalomyosynangiosis, and one case was treated with direct revascularization surgery with anastomosis of the superficial temporal artery-middle meningeal artery (STA-MCA).

50% of the cases did not present neurological deficit or alteration after the proposed treatment, of the other 50%, the most observed sequelae were hemiparesis, hemiplegia, and paralysis of the seventh pair. There was no associated mortality described in the study.

Discussion

MS has been described mainly in Asia; however, its incidence has been growing in America and Europe. In the literature, a predominant disease is found in the female sex; within the present study, an equal distribution of sexes was found. The most frequent initial manifestations were two cases of seizure episode and two cases of intense headache, one patient presented neurological deficit due to hemiplegia, one case drowsiness and another case loss of consciousness; of the above, ischemic stroke was associated with cases that

debuted with hemiplegia, somnolence, and hemorrhagic stroke to cases of seizure and headache, this is compatible with the literature where the main initial manifestations are described as strokes, either ischemic or hemorrhagic in both adults and children. As a standard diagnostic study, cerebral panangiography was performed in all six cases, all with findings compatible with MS, mainly bilateral stenosis of internal carotid arteries in their terminal portion. Although panangiography is the GS diagnostic method (Fig. 1), in three of the cases, other types of images were previously performed, such as brain resonance imaging and cerebral resonance angiography, where the same characteristics mentioned (carotid stenosis) were observed. The Suzuki classification was used in the diagnostic examination, with four Suzuki V patients predominating. The treatment of choice was surgical in most cases, with indirect revascularization predominating in 5 of them.

Finally, at follow-up, 3 cases with neurological sequelae were identified within these paresis of the VII pair, right superior hemiparesis and left hemiplegia, no case of mortality associated with management or moyamoya disease was described.

Conclusion

MS or MS, as the case may be, speaks of a progressive occlusion of the cerebral arteries, mainly the internal carotid arteries, presenting as a compensatory method, the formation of a collateral circulation network, weak, prone to hemorrhages or ischemia, the latter frequently occurring in children. The definitive diagnostic method is cerebral panangiography where the vessels are observed in "cigarette smoke" highly compatible with moyamoya. Within our experience in the institution, all cases were approached in the same way, in all of them, arteriography was performed and later surgical management of revascularization, in the early follow-up, three cases were observed with neurological deficit which may be associated with the ischemic event presented on admission, five of the six patients were discharged with a Glasgow score of 15/15 and only one patient 14/15 due to disorientation, after 1 month, arteriographic control was performed in 3 cases with adequate neovascularization and anastomosis, in three of the remaining cases, no control was performed. It is pertinent to continue long-term follow-up to establish sequelae and possible recurrences of late presentation. Studies in our country are limited and research on this disease/syndrome should continue in an institutional and multicenter manner as far as possible to clarify improvements in treatments, diagnoses, and follow-up of patients.

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Conflicts of interest

The authors declare no conflicts of interest.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients or subjects mentioned in the article. The corresponding author is in possession of this document.

Use of artificial intelligence for generating text. The authors declare that they have not used any type of generative artificial intelligence for the writing of this manuscript, nor for the creation of images, graphics, tables, or their corresponding captions.

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