Case series: Moyamoya syndrome in patients with Down syndrome at the Instituto Nacional de Pediatría

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BACKGROUND

Moyamoya disease is a progressive stenosis of intracranial arteries that promotes the development of collateral vessels. When associated with conditions such as Down syndrome (DS) or autoimmune disorders, it is termed Moyamoya syndrome (MMS). MMS can manifest as transient ischemic events, neurological deficits, or remain asymptomatic. the prevalence reported in the literature in the general population ranges from 0.8-16.1/100,000, whereas in patients with DS it reaches 3,800/100,000.

OBJECTIVE

To describe the clinical characteristics, Regarding management, outcomes, and APS testing results in patients with Down syndrome and Moyamoya syndrome.

METHODS

Study design: retrospective case series
Inclusion criteria: Patients diagnosed with both Down syndrome and Moyamoya síndrome treated at the National Institute of Pediatrics, ranging in age from 1 day to 17 years and 11 months.
Exclusion criteria: Patients older than 18 years, patients in whom the diagnosis of Moyamoya síndrome was not confirmed.
Study description: A retrospective case series study was conducted using a database created from electronic records of patients with Down syndrome and Moyamoya syndrome from the Instituto Nacional de Pediatría from the period 2015-2025, taking into account sex, age of the patient at diagnosis of MMS, presence of symptoms, presence of ischemic stroke or intracranial hemorrhage, diagnostic method, APS testing results, treatment and follow up duration.

RESULTS

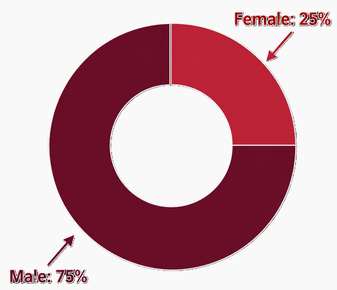

Figure 1. Patient demographics by sex

The study reviewed 8 patient records of individuals with Down syndrome diagnosed with Moyamoya syndrome. Most patients were male (75%). The patients' ages ranged from 2 to 9 years, with a predominance of younger children. The most frequent age was 2 years, representing 62.5% of cases (5 patients), while the remaining ages (5, 7, and 9 years) each accounted for 12.5% (1 patient each).

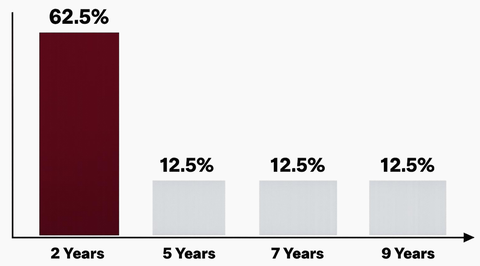

Figure 2. Patient demographics by age

Regarding symptoms, The most frequent manifestation was hemiparesis and ischemic stroke in 87.5% of the patients, followed by epileptic seizures in 62.5%, and less frequently, intracranial hemorrhage in 12.5%. Only 12.5% presented with visual changes.

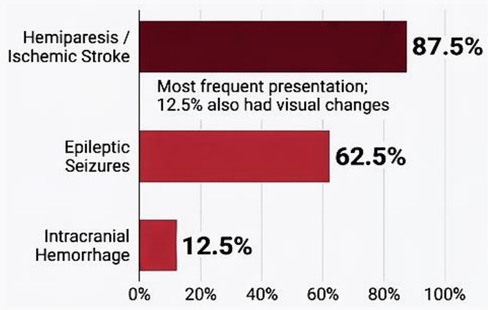

Figure 3. Clinical manifestations

Regarding diagnosis, angiography was used in 75% of cases, making it the most frequently used diagnostic method. Magnetic resonance imaging (MRI) was performed in 62.5% of cases, computed tomography (CT) in 50%, electroencephalography (EEG) in 25%, and SPECT in only 12.5%.
With respect to comorbidities associated with Down syndrome, 50% of patients presented with congenital heart disease, 37.5% with epilepsy, and 12.5% with hypothyroidism, visual impairment, or hearing loss.
As for antiphospholipid testing status, 50% tested positive and 25% tested negative or was not performed.

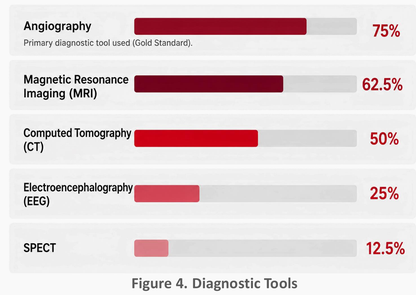

Figure 4. Diagnostic Tools

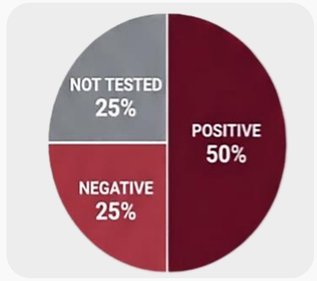

Figure 5. APS testing status

Talking about treatment, 62.5% received conservative management with aspirin, 25% underwent surgical treatment, and 12.5% remained under observation.

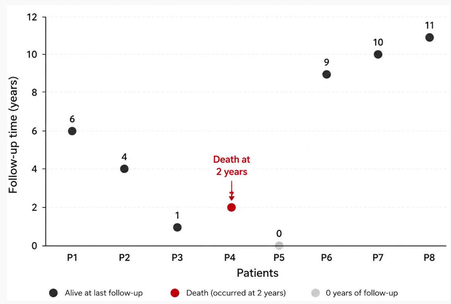

Figure 6. Survival outlook

The mean follow-up duration after Moyamoya diagnosis was 5.86 ± 4.38 years (median: 6 years; range: 0–11 years; n=7). One death was recorded occurring 2 years after diagnosis.

CONCLUSIONS

Moyamoya syndrome in patients with Down syndrome presents predominantly at a young age and manifests primarily with ischemic cerebrovascular events and hemiparesis. Angiography remains the most widely used diagnostic method, and management is usually conservative in most cases. The presence of antiphospholipid antibodies in a significant proportion of patients suggests a possible role in the pathophysiology or as a contributing factor to thrombotic risk; therefore, their evaluation could be relevant in the comprehensive management of these patients.
Limitations: The retrospective, single-center design limits the generalizability of the results. In addition, referral-center recruitment may have introduced selection bias, with a possible overrepresentation of more severe cases. Therefore, larger multicenter studies are needed to better characterize the relationship between Down syndrome, Moyamoya syndrome, and the presence of antiphospholipid antibodies.

REFERENCES

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