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IMAGES IN CLINICAL NEUROLOGY

A rare cause of stroke: Coexistence of Down syndrome and Moyamoya disease

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Moyamoya disease (MMD), a rare cerebral circulation anomaly, is a chronic condition characterized by the development of multiple collateral anastomoses between the internal carotid artery and the external carotid artery as a result of occlusion and stenosis in the distal regions of the carotid arteries, particularly in the proximal segments of the anterior and middle cerebral arteries.[1] Although it is typically observed bilaterally, it may develop unilaterally in 20% of cases. The disease derives its name, Moyamoya, from a Japanese term referring to the "puff of smoke" appearance of the collateral anastomoses in the anterior cerebral circulation observed in angiographic imaging. The first case was reported in 1957, and the def§inition of the disease was established in 1969. The association of MMD with conditions such as Graves' disease, Behçet's disease, antiphospholipid antibody syndrome, polycystic kidney disease, neurofibromatosis, and Down syndrome is referred to as Moyamoya syndrome.[2] The relationship between MMD and Down syndrome has been debated in rare reported cases. Herein, we presented the radiological images of a patient with Down syndrome who was diagnosed with Moyamoya syndrome, highlighting the rare coexistence of these two conditions.

An 18-year-old female patient was admitted to the neurology outpatient clinic due to a sensation of numbness in the right hand lasting less than 24 h and staring spells over the past week. The patient, diagnosed with Down syndrome, had a history of patent ductus arteriosus closure surgery performed in 2011. Physical examination revealed stigmata of Down syndrome. The patient exhibited a reduced interest in her surroundings that was inconsistent with her age, while the somatic neurological examination was normal. Laboratory tests revealed antinuclear antibody positivity at a 1:100 dilution. Magnetic resonance angiography of the brain showed a thin appearance of the right internal carotid artery (ICA) and severe stenosis in the terminal regions of both ICAs. Multiple small vascular structures branching from the terminal regions of the ICAs, representing collateral anastomoses, were observed (Figures 1 and 2).

Patients with Down syndrome are at a higher risk of thromboembolic ischemic stroke due to associated cardiac malformations. Additionally, increased endostatin concentrations in these patients are considered a predisposing factor for the development of abnormal vascular networks.[3] Furthermore, it was suggested that, as in Graves' disease, the overexpression of systemic and endothelial inflammation markers and immune dysregulation in patients with Down syndrome play a role in the etiology of MMD.[3,4] While mutations in regions such as 3p24-26, 6q25, 8q23, 10q23.31, 12p12, and 17q25 were implicated in the pathogenesis of MMD, and 10% of cases were associated with genetic factors, a genetic determinant linked to chromosome 21 has not been identified.[5] Proteins encoded on chromosome 21, such as collagen type VI alpha chain, cystathionine

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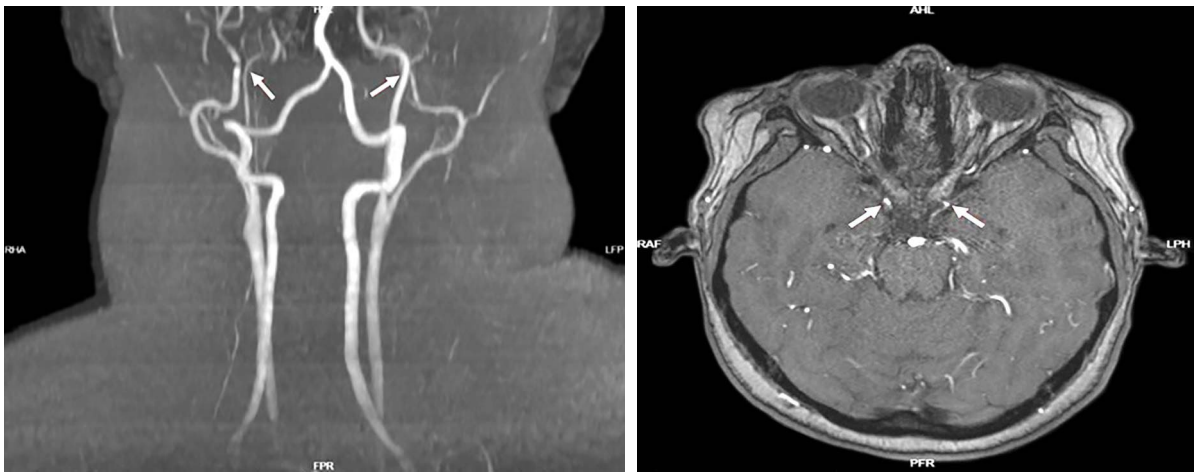

Figure 1. White arrows indicate severe stenosis in the apical regions of the bilateral internal carotid arteries.

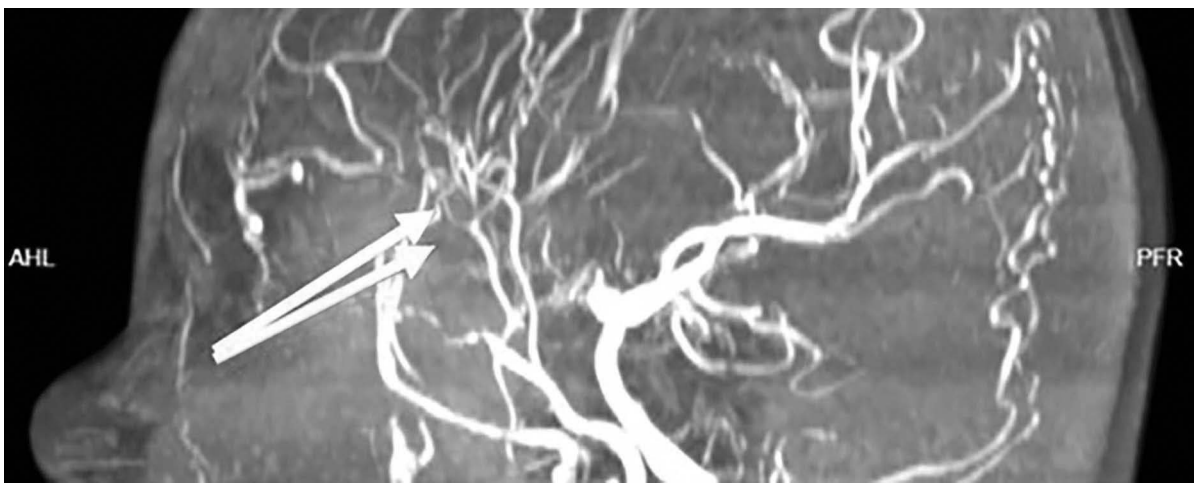

Figure 2. Multiple small vascular structures branching from the apical region of the internal carotid arteries, consistent with Moyamoya; collateral anastomoses are marked with white arrows.

beta-synthase, and interferon-gamma receptor, are thought to be associated with a high risk of vascular diseases and may indirectly play a role in the etiology of MMD.[2]

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

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