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Moyamoya Syndrome Associated With Down Syndrome: A Case Report With A Literature Review

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Abstract :
Moyamoya syndrome is rare. It is important to differentiate between the primary form of this syndrome and the secondary form, which may be linked to certain conditions such as neurofibromatosis, homocystinuria, and trisomy 21. We recently observed a 23-year-old patient with trisomy 21 who exhibited motor deficits in all four limbs. Initial brain imaging revealed multiple bilateral ischemic strokes of varying ages. The diagnosis of Moyamoya syndrome was based on the presence of stenoses in the M1 segment of the right and left middle cerebral arteries. Blood tests indicated hyperthyroidism along with elevated anti-TPO and anti-THSH antibodies. The patient was treated with antiplatelet therapy, synthetic antithyroid drugs, and physiotherapy sessions, and showed partial recovery of the motor deficit over time.

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I. Introduction :

Moya-Moya syndrome or disease is used to define the presence of an abnormal vascular network, developing as a result of progressive stenosis (narrowing) of the arteries located at the base of the brain (termination of the internal carotid artery and beginning of its branches: anterior cerebral artery and middle cerebral artery). This abnormal vascular network appears progressively to compensate for reduced blood flow downstream of stenoses in the arteries at the base of the skull. It often takes on a "cloudy" or "smoke-like" appearance, which translates into Japanese as "Moya-Moya". This abnormal vascular network can develop when narrowing of the arteries at the base of the skull occurs as part of a known general or local disease. This is known as "Moya-Moya" syndrome. When no disease is associated with the arterial anomalies, the term "Moya-Moya" disease is preferred [1].

II. Observation :

A 23-year-old patient with trisomy 21 presented one month before admission with right-hemisphere weakness and speech suspension. Symptomatology worsened 3 weeks later with the appearance of a left-hemisphere motor deficit. Clinical examination on admission found the patient conscious, normotensive, tachycardic at 114, and apyretic. Neurological examination revealed bilateral pyramidal deficit syndrome and aphasia. Cerebral MRI showed recent ischemic lesions in the territories of the right and left superficial sylvian arteries visible in diffusion hypersignal with ADC restriction and old sequelae visible in FLAIR sequence. The TOF sequence showed slowed flow in the M1 segments of both right and left middle cerebral arteries, consistent with stenoses suggestive of MOYA MOYA syndrome. The ECG showed sinus tachycardia, and the transthoracic ultrasound performed as part of the etiological work-up returned normal. Biological workup revealed hyperthyroidism with TSH at 0.017 IU/ml associated with elevated anti-TPO antibodies at 254 IU/ml and anti-TsH antibodies at 25 IU/ml. Cervical ultrasound showed a moderate increase in thyroid size. These findings were consistent with Graves' disease. The patient was treated by antiplatelet therapy, synthetic antithyroid drugs and physiotherapy. After 3 months, the motor deficit improved.

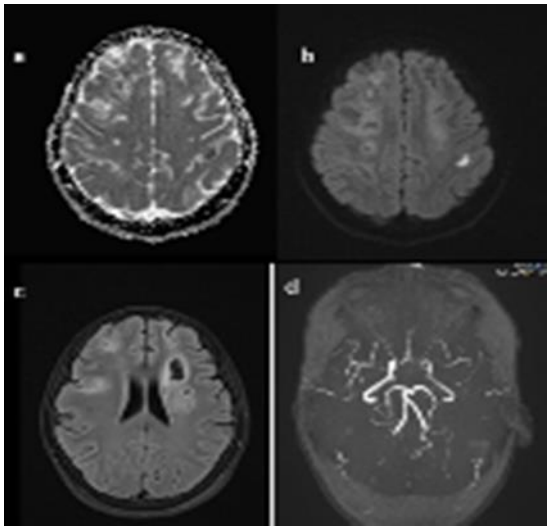

Figure 1: Cerebral MRI showed recent ischemic lesions in the territories of the right and left superficial sylvian arteries visible in diffusion hypersignal (b) with ADC restriction (a) and old sequelae visible in FLAIR sequence. The TOF sequence (d) showed slowed flow in the M1 segments of both right and left middle cerebral arteries.

III. Discussion :

Down syndrome is a highly common chromosomal defect in approximately one out of every 800 live births [2]. Down syndrome is primarily caused by chromosome 21 trisomy. Individuals with Down syndrome now have a life expectancy of roughly 60 years [3]. For a variety of reasons, including vascular abnormalities and congenital heart disease, patients with Down syndrome are vulnerable to cerebrovascular accidents. Moyamoya disease is uncommon, progressive stenosis of many cerebral arteries [4]. As a result of this blockage, a vascular network grows around the stenosed vessel. Collaterals are small and delicate vessels that are prone to bleeding, aneurism, or thrombus formation. On standard angiographic imaging, this vascular network appears as a "puff of smoke" (referred to in Japanese as the moyamoya phenomenon) [3]. Moyamoya is classified into two subtypes: idiopathic moyamoya illness and moyamoya syndrome. The vascular alterations in moyamoya syndrome are frequently associated with other syndromes or systemic disorders, examples include Down syndrome, sickle cell anemia, neurofibromatosis type-1, congenital heart disease, fibromuscular dysplasia, activated protein C resistance, and head trauma [5-6]. Moyamoya syndrome in trisomy 21 might manifest clinically as transitory ischemia symptoms or as a neurological disability. Other people may present with no symptoms, and these distinctive vascular alterations may be discovered by chance. Typically, standard angiographic imaging is used to confirm the diagnosis. Moyamoya syndrome in trisomy 21 was poorly understood in terms of its origin and pathophysiology. This comprehensive review attempts to gain a better understanding of the disease's pathophysiology in Down's syndrome.

IV. Conclusion :

The knowledge regarding the association of MMD in a patient with DS with stroke is important so as to reach a correct diagnosis and establish an appropriate management plan. The reason of this association is still not clear. The evaluation must include an MRA. Further advanced imaging, autoimmune work-up, or epigenetic studies are needed to find out underlying basis for increased incidence of MMD in trisomy 21 cases.

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