Beta-thalassemia intermedia associated with moyamoya syndrome

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Abstract
Moyamoya syndrome (MMS) is a progressive disorder. We report a 19-year-old boy with β-thalassemia who presented with a left hemiparesis. Brain MRI showed old middle cerebral artery and left frontal subcortical white matter infarcts. Brain magnetic resonance angiography and digital subtraction angiography revealed occlusion of the bilateral internal carotid arteries with a rich network of basal collateral vessels. To our knowledge this is the third report of β-thalassemia intermedia and MMS, and the first report of a patient in Turkey. It emphasizes the potential for cerebral infarct due to anemia, protein S and thrombocytosis.

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1. Introduction
Moyamoya syndrome (MMS) is characterized by typical angiographic “moyamoya” changes associated with subsequent clinical features.1,2 In thalassemia intermedia (TI), a low level of hemoglobin causes hypoxia, hypertrophic changes of vascular endothelium, and microvascular stenosis.3–5

2. Case report
A 19-year-old boy, who had difficulty walking, underwent a neurological examination. He started to have walking difficulties at the age of 5 years, and which had gradually deteriorated in the 3 years prior to presentation. At 1 year of age, he had been diagnosed with TI, and had undergone a splenectomy at 17 years of age.

Physical examination revealed pale and icteric skin, small stature and a systolic murmur at the cardiac apex. Neurological examination showed a left hemiparesis with power of 4/5 in his left arm and lower extremity, bilateral brisk deep tendon reflexes, and a bilateral positive Babinski sign. The results of peripheral blood analysis were: erythrocytes, 3.3 × 1012/mm3; hemoglobin, 8.09 g/dL; mean corpuscular volume, 74.5 fL; platelet count, 1.180 × 1012/mm3. Target cells showed anisocytosis, poikilocytosis, and hypochromic microcytic anemia. Hemoglobin (Hb) electrophoresis revealed the following: Hb A, 64.2%; Hb F, 32.4%; and Hb A2, 6.4%. Blood chemistry tests and coagulation studies revealed normal protein C, antithrombin III, prothrombin time and low protein S. Electrocardiography was normal. Brain MRI and magnetic resonance angiography revealed chronic infarcts at the right fronto-temporo-parietal region, nucleus lentiformis, thalamus and at the left frontal subcortical white matter accompanied by moyamoya vessels in the lenticulostriate and thalamoperforate distributions, and occlusion of both internal carotid arteries at their origin. Digital subtraction angiography revealed narrowing of both internal carotid arteries. The artery terminated at the intracranial segment and, thus, normal cerebral arteries could not be discerned; there

Fig. 1. (a) The fluid attenuated inversion recovery axial brain MRI showing a right fronto-temporo-parietal and a left frontal infarct. (b) Digital subtraction angiogram. Anteroposterior projection of the left common carotid artery injection showing prominent branches from the external carotid artery, and irregular very thin collaterals branching from the internal carotid artery (the “moyamoya” or “puff of smoke” vessels).

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were irregular collateral arteries instead (Fig. 1a and b). The left vertebral artery was narrow, and the basilar artery and posterior cerebral arteries were the major feeders of vascular brain parenchyma with irregular collateral arteries.

3. Discussion

Although stroke is a serious complication of thalassemia major (TM), it is only reported with an annual incidence of 0.1% to 0.3% per year.3 There have been 2 reports of the association of moyamoya vasculopathy with β-thalassemia: (i) a 5-year-old girl with β-TM who presented with hemiconvulsions and hemiplegia with cerebral infarction and with moyamoya vessels, and who consequently suffered from aphasia and spastic paraplegia;46 and (ii) a 14-year-old girl with β-TI, who developed MMS after splenectomy and experienced transient ischemic attacks.2 Our patient had spastic left hemiparesis and pyramidal signs on both sides. Although he had experienced the symptoms for 14 years, he had not consulted a neurologist. Cranial MRI demonstrated chronic right fronto-temporal-right parietal and left fronto-temporal infarcts, some of which were symptomatic and some were silent. These findings suggested that multiple cerebral infarcts and large arterial occlusions were related to his chronic hypercoagulable state. The risk of a hypercoagulable state in thalassemia is multifactorial (including altered platelet function, endothelial activation, red blood cell membrane abnormalities leading to activation of the coagulation cascades, and changes in coagulation protein levels).

In conclusion, TI may have a role in the etiology of moyamoya vasculopathy. Accordingly, patients with TI should be monitored closely for stroke. Stroke can be associated with moyamoya vessels and silent progression in patients with TI.

References


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Extraventricular neurocytoma with atypical features and ganglionic differentiation

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Extraventricular neurocytoma (E VN) is an unusual variant of central neurocytoma located outside the ventricular system. Both tumours share a similar histology characterised by monotonous populations of round-to-oval cells with scanty cytoplasm separated by neuropil and branching capillaries. We report an EVN arising from the right frontal lobe near the olfactory tract in a 34-year-old male with worsening chronic epilepsy. Our patient’s tumour exhibited many uncommon features including ganglionic differentiation, increased mitotic activity and a high proliferative index. We discuss the important differential diagnoses given the site of the tumour as well as the differentiating features from olfactory neuroblastoma, oligodendroglioma, anaplastic ganglioglioma and supra-tentorial primitive neuroectodermal tumour.

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1. Case report

The patient was a 34-year-old male with a long history of epilepsy from a right gyrus rectus lesion. The frequency of his seizures had increased in the months prior to surgery despite an increased dose of anti-convulsant therapy.

An MRI showed an ill-defined 18 mm × 21 mm × 21 mm lesion in the right frontal lobe. The lesion had a low signal on a T1-weighted MRI and was hyperintense on fluid-attenuated inversion recovery with nodular enhancement in its antero-medial aspect after gadolinium enhancement (Fig. 1). No increase in uptake was detected by positron emission tomography.

A stereotactic right frontotemporal craniotomy was performed. The tumour was located in the right gyrus rectus of the frontal lobe and adhered to the olfactory tract. Complete macroscopic resection of the tumour was achieved, and confirmed by the image guidance system.

Histological sections showed fragments of brain tissue infiltrated by tumour cells with round-to-oval nuclei and scanty cytoplasm. They were embedded within a fine neuropil background. The tumour cells were arranged in sheets, ribbons or nodules separated by a fine vascular network. There was mild-to-moderate nuclear pleomorphism and focal anaplasia with increased mitotic