MOYA MOYA DISEASE IN A PATIENT OF THALASSEMIA MAJOR - CASE REPORT

Radiodiagnosis

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ABSTRACT

The moyamoya syndrome is a cerebrovascular condition that predisposes affected patients to stroke due to progressive stenosis of the intracranial internal carotid arteries and their proximal branches. It can be seen in association with several inherited disorders. However, its association with thalassemia is very rare.

KEYWORDS

MRI, thalassemia, Moya Moya syndrome

INTRODUCTION:

The moyamoya syndrome is a cerebrovascular condition that predisposes affected patients to stroke due to progressive stenosis of the intracranial internal carotid arteries and their proximal branches. Reduced blood flow in the major vessels of the anterior circulation of the brain leads to compensatory development of collateral vasculature by small vessels near the apex of the carotid, on the cortical surface, leptomeninges, and branches of the external carotid artery supplying the dura and the base of the skull. In rare cases, this process also involves the posterior circulation, including the basilar and posterior cerebral arteries. [1]

The word "moyamoya" means "puff of smoke" in Japanese, a term describing the appearance of this cluster of tiny blood vessels.

Ischemic attacks of hemiplegia or seizures constitute the major presentation in childhood, whereas subarachnoid hemorrhage occurs more frequently in adults. The progressive nature of the disease leads to unfavorable neurologic outcomes without surgical intervention to reconstitute the cerebral circulation. [2]

Case Presentation:

A 13-year female patient diagnosed as thalassemia major, complaining of right upper limb spastic posture with flexion of hand associated with weakness of right hand. Patient had past history of multiple blood transfusions. No other neurological complaints were present as seizure or headache. On examination: weakness of right upper limb was present. Power in left upper limb and bilateral lower limb was normal. MRI Protocol: Multiplanar multiecho MRI of the brain was performed on Philips Achieva 3T MRI. In addition, MR angiogram of brain was performed using Time of Flight (TOF) sequences.

MRI findings included:

- Chronic infarct in the left fronto-parietal cortex and parafalcine right frontal cortex.
- Thickened skull vault likely related to the extramedullary hematopoiesis- thalassemia.
- The choroid plexuses in bilateral lateral and fourth ventricles appeared prominent.
- Moderate to severe narrowing of the ICA in the petrous and cavernous segments with severe stenosis of the supra-clinoid ICA.
- Severe stenosis of right A1, left A1, left M1 and M2 with multiple collateral vessels along bilateral MCA and left ACA (puff of smoke appearance)

Figure 1 a,b,c: AXIAL FLAIR images showing chronic infarct in left fronto-parietal region

Figure 2: T1 Sagittal images: chronic infarct in left frontoparietal region

Figure 3: T1 Sagittal image : chronic infarct in right frontal parafalcine region

Figure 4: T2 coronal image: chronic infarct in left frontoparietal region

Figure 5: T2 coronal image: chronic infarct in right frontal parafalcine region and left fronto-parietal region.

Figure 6 a, b: GRE sequences showing foci of blooming in bilateral lateral ventricle, IVth ventricle and foramen of Luschka – related to prominence of choroid plexuses.

Figure 7 a, b, c: 3D angiography axial section: show moderate to severely narrowing in petrous and cavernous segment with severe stenosis of supraclinoid segment of left ICA

Figure 8 a, b: 3D angiography volume images and axial images: show multiple collateral vessels noted along bilateral MCA (R>L) appearing puff of smoke appearance.

Figure 9 a, b: Mild narrowing of the right cavernous ICA and right MCA with severe narrowing at supraclinoid ICA after ophthalmic artery. The basilar artery and bilateral vertebral arteries appear prominent. These features are s/o secondary Moyamoya disease.

DISCUSSION:

Moyamoya disease is a cerebrovascular condition predisposing affected patients to stroke in association with progressive stenosis of the intracranial internal carotid arteries and their proximal branches. Patients with characteristic moyamoya vasculopathy plus associated conditions are categorized as having moyamoya syndrome[4].

Moyamoya syndrome occurs in patients with brain tumors, vascular

malformations, or vasculitis after irradiation, infections, and head trauma. It can be associated with various inherited disorders, that is, neurofibromatosis, tuberous sclerosis complex, Down syndrome, and Alagille syndrome, along with some anemias[5]. Patients with homozygous hemoglobin S occasionally develop cerebral infarctions and narrowing or occlusion of the internal carotid arteries, with the moyamoya pattern as the most common arteriographic finding[6,7]. The progressive vasculopathy can be a consequence of the flow in the vasa vasorum being obstructed by sickle cells, which leads to ischemia in the walls of the internal carotid arteries, intimal proliferation, and occlusion but not exclusively of the direct obstruction of small cerebral vessels by the cells. Moyamoya syndrome has been reported in patients with hereditary spherocytosis[8] and Fanconi anemia[9]. Ischemic strokes might be associated with other types of anemia, including iron deficiency anemia and Diamond-Blackfan anemia[10]. There is a high incidence of cerebral thrombosis in alpha and beta-spectrin-deficient mice with hereditary spherocytosis[11]. The increased blood flow caused by anemia, together with less deformable red cells, can lead to progressive endothelial proliferation and subsequent vascular occlusion.

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)[12].

The beta-thalassemias are a heterogeneous group of inherited hemoglobin disorders characterized by reduced synthesis of beta-globin chains and primarily affecting individuals in malaria-endemic areas[13]. Homozygous or compound heterozygous mutations of the beta-globin gene lead to severe anemia with ineffective erythropoiesis and hemolysis as the major or intermediate phenotype.

An underlying chronic hypercoagulable state resulting in thrombotic events has been well described in beta-thalassemia. The risk factors described are older age, severity of anemia, lack of regular transfusions, splenectomy, and a family history of thrombotic events[14,15]. The imbalance resulting in the procoagulant state has been described[16]. The abnormal findings detected include low levels of proteins C and S, enhanced platelet consumption, ongoing platelet, mono-cyte, granulocyte, and endothelial activation, increased cohesiveness of red blood cells (RBCs) to epithelial cells, and increased negatively charged phospholipids on RBCs.

There have been very few case reports (less than 10) of cases with thalassemia and Moyamoya disease.

Teaching point:

Moyamoya disease can be present seen in patient suffering from thalassemia as in our case. MRI is very helpful in such case, apart from vasculopathy MRI also reveals grey white matter changes, various ischemic changes, infarction, brain atrophy and ventricular dilatation and MR imaging plays an important role in the diagnosis of moyamoya disease

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