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Moya Moya syndrome in a HbE beta thalassemic child presenting with hemiplegia in rural West Bengal

Dr Kashi Nath Sarkar[1], Dr Debasis Deoghuria[2], Dr Manisha Sarkar[3], Dr Shivani Sarkar[4]

[1](Junior Resident, Department of Radio diagnosis, Bankura Sammilani Medical college, Bankura, India)
[2](Associate Professor, Department of Radio diagnosis, Bankura Sammilani Medical college, Bankura, India)
[3](Junior Resident III, Department of Community Medicine, Medical College, Kolkata, India)
[4](Junior Resident II, Department of Pathology, NRS Medical College, Kolkata, India)

Abstract : *Moya Moya disease (primary) is an idiopathic, non inflammatory, non-atherosclerotic, progressive vasculo-occlusive disease involving the circle of Willis, typically the supraclinoid internal carotid arteries, followed by extensive collateralization which are prone to hemorrhage, aneurysm and thrombosis. Secondary Moya Moya phenomenon or Moya Moya syndrome occurs in a wide range of clinical scenarios including prothrombotic states such as sickle cell anemia, however it has rarely been reported in association with HbE beta thalassemia intermidia. Here we are reporting Moya Moya syndrome in a HbE beta thalassemic child presenting with transient ischemic stroke and hemiplegia, primarily to emphasize thalassemia as an extremely rare but potential etiology of Moya Moya syndrome and that children with thalassemia should be screened for future risk of Moya Moya syndrome and transient ischemic stroke. The disease is progressive and an early radiological diagnosis followed by prompt surgical revascularization remains the only hope for improved neurological prognosis.*

Keywords - *Moya Moya syndrome, HbE beta thalassemia intermidia, rare, transient ischemic stroke, hemiplegia*

I. Introduction

The term Moya Moya literally is Japanese expression for "hazy, just like a puff of cigarette smoke drifting in the air" and was first described by Suzuki and Takaku in 1969 enchanting the angiographic appearance of dilated and irregular collateral vessels that develop secondary to progressive occlusion of the internal carotid arteries and circle of willis.[1,2] Moya Moya disease is an idiopathic, non-inflammatory, non-atherosclerotic, progressive vasculo-occlusive disease involving the circle of Willis, typically the supraclinoid internal carotid arteries, followed by anterior cerebral and middle cerebral arteries. [1-3] Primary Moya Moya disease is idiopathic and has been reported principally in Japan.[3] Secondary Moya Moya (Moya Moya syndrome or Moya Moya phenomenon) occurs in association with other disorders such as neurofibromatosis type 1, tuberous sclerosis, systemic lupus erythematosus, antiphospholipid antibody syndrome, infective etiologies viz, tuberculous meningitis, bacterial meningitis, down syndrome, aplastic anemia, prothrombotic states like sickle cell anemia.[1,3-5] Although Moya Moya syndrome is the most common cause for transient ischemic stroke in asian children,[4] it has been on extremely rare cases demonstrated in association to thalassemia intermidia.[1,4,6] In our case we report a HbE beta thalassemic child presenting with hemiplegia, who on magnetic resonance angiography showed features of Moya Moya disease i.e. flow voids in basal ganglia and "Puff of smoke" like lenticulostriate and thalamostriate collaterals on angiography.[1,4] All other common associations of Moya Moya disease were ruled out.

II. Case Report

An 11 year old male child reported to radio diagnosis department for MRI with history of recurrent and progressive hemiplegia. On taking history and detailed clinical evaluation we found out that the child had multiple episodes of syncope, sudden fall while playing, which had gradually deteriorated prior to presentation with right sided hemiplegia for last 1 year, left sided hemiplegia for last 1 month. There was no history of fever, convulsion, head injury, ear discharge and delayed mile stones. There were no signs of meningeal irritation. On examination child was found to be conscious but disoriented. The child had hemiplegic gait, decreased right side tone, muscle power grade of 2 (muscle contraction was seen only when force of gravity was eliminated) over right upper and lower limb, bilateral brisk deep tendon reflexes and bilateral positive Babinski sign. There were no neurocutaneous nodules and facial asymmetry. All other systems were found to be normal. The child was diagnosed with HbE beta thalassemia intermidia at 5 years of age and had received multiple blood transfusions thereafter. The child had short stature, icteric, pale and anemic skin. MRI and MRA findings revealed it to be a

case Moya Moya syndrome. MRI showed encephalomalacia in bilateral cerebral hemisphere (Left>Right), Old infarct of left basal ganglia and periventricular white matter. We tested the child on hematological, biochemical and serological parameters(as tabulated below) for all common secondary associations of Moya Moya syndrome, however found a very rare entity i.e. HbE beta thalassemia as its primary etiological association.

Fig 1 shows a child with right sided Hemiplegia

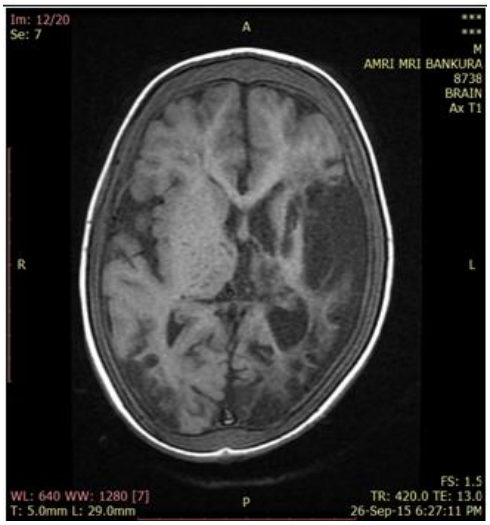

Fig 2 Axial T1WI taken at TR 420ms and TE 13ms shows multiple punctate signal flow voids in bilateral basal ganglia representing lenticulostriate and thalamostriate arterial collaterals[4]

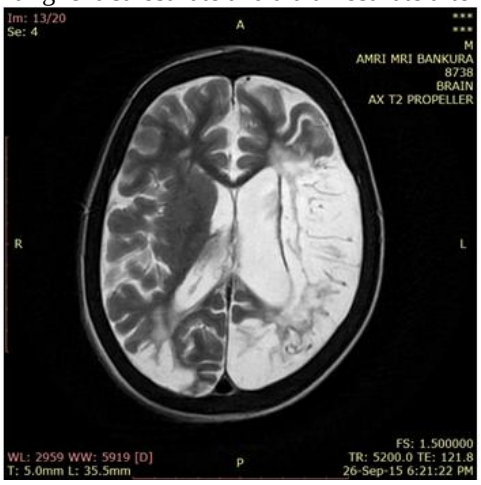

Fig 3 Axial T2WI sequences taken at TR 5200ms and TE 121ms shows net like cisternal filling defects[3,4]

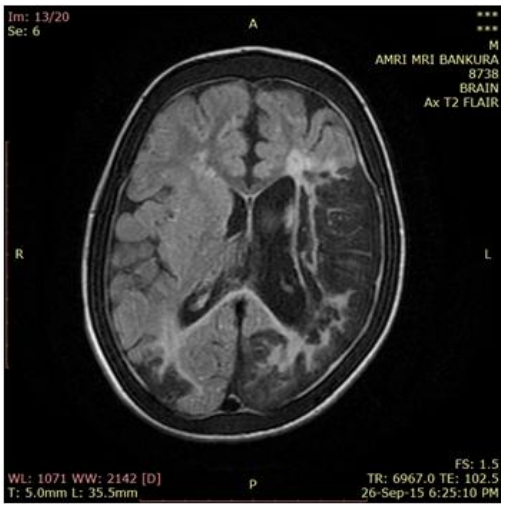

Fig 4 Axial T2WI FLAIR sequences taken at TR 6967ms and TE 102ms shows encephalomalacia in bilateral cerebral hemisphere (L>R) and hyper intense cortical sulci i.e. 'Leptomeningeal "Ivy Sign" due to slow flow in engorged pial collaterals [1,4]

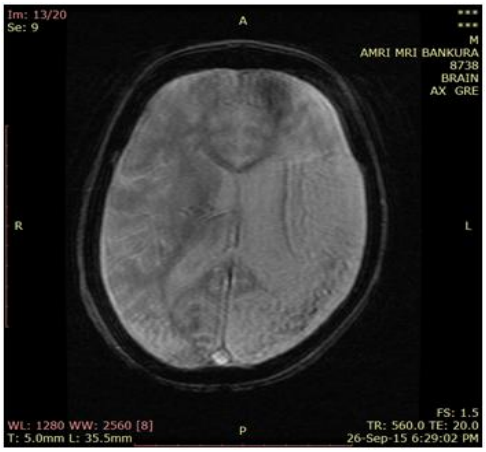

Fig 5 Axial GRE taken at TR 560ms, TE 20ms and 15.7 khz frequency shows no features of hemorrhage and calcifications[1,4]

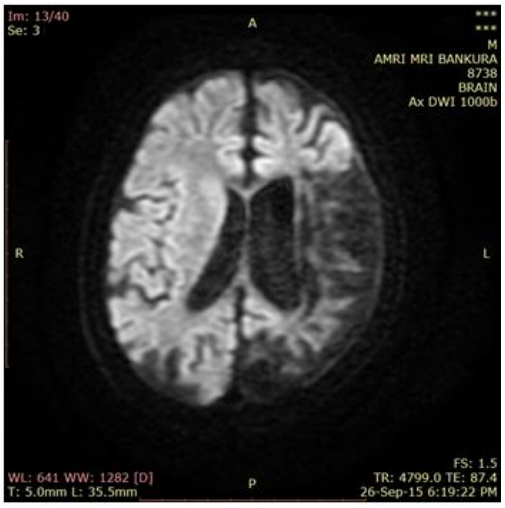

Fig 6 Axial DWI taken at TR 4799ms, TE 87.4ms and 250 kHz shows chronic left Para ventricular white matter infarct[1,3,4]

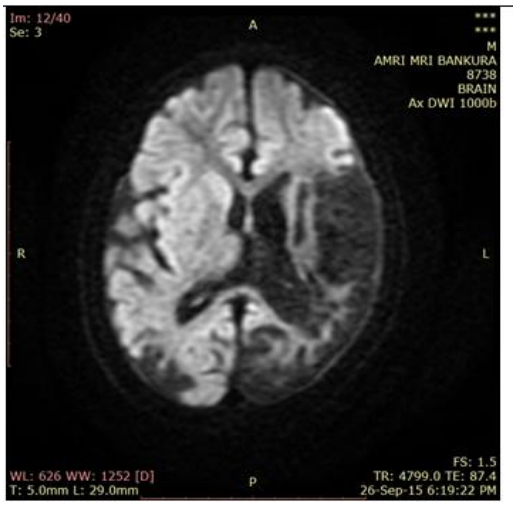

Fig 7 Axial DWI taken at TR 4799ms ,TE 87.4ms and 250 kHz shows chronic left basal ganglia and Para ventricular white matter infarct [1,3,4]

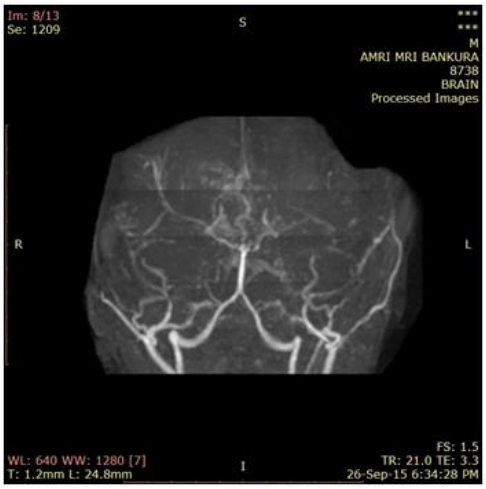

Fig 8 MR Angiography shows narrowed bilateral internal carotid , anterior and middle cerebral arteries with extensive lenticulostriate collaterals giving "puff of cigarette smoke" appearance[1,4]

III. Figures And Tables

TEST DESCRIPTION	**OBSERVED VALUES**	**REFERENCE RANGE AND UNITS**
Red Blood Cell Count (RBC)	5.95 million/µL/cu mm	4.2 - 6.9 million/µL/cu mm
WBC (white blood cell count)	10.5 × 103/mm3	4.3-10.8 × 103/mm3
WBC Differential leukocyte count	Segmented Neutrophils70 % Lymphocytes 23 % Monocytes 02 % Band neutrophils 01 % Eosinophils 05 % Basophils 00%	Segmented Neutrophils 45 – 75% Lymphocytes 16 – 46% Monocytes4 – 11% Band neutrophils 0 – 5% Eosinophils 0 – 8% Basophils 0 – 3%
Erythrocyte Sedimentation Rate (ESR)	60 mm /hr	Male: 1 - 13 mm/hr, Female: 1 - 20 mm/hr
Reticulocytes	2.1%	0.5%-1.5%
Hemoglobin	12.5gm/dl	Male: 13 - 18 gm/dL
Packed cell volume (PCV)	46.6 %	Male 42-52%
Mean Corpuscular Volume (MCV)	75.3 cu µm	76 - 100 cu µm
Mean Corpuscular Hemoglobin (MCH)	24.4 pg/cell	27 - 32 pg/cell
Mean Corpuscular Hemoglobin Concentration (MCHC)	31.1%	32 - 36% hemoglobin/cell
Red blood cell distribution width (RDW)	14.9%	11.5%-14.5%
Platelet Count	186,000 /mL	150,000 - 350,000/mL
Complete blood picture	Anisopoikilocytosis with predominantly microcytic hypochromic RBCs. Few target cells & tear drops cells also seen.	

TEST DESCRIPTION	OBSERVED VALUES	REFERENCE RANGE AND UNITS
Hb electrophoresis		
Hemoglobin A	11.40	94.3-98.5%
Hemoglobin F	20.2	00-02%
Hemoglobin A2+ Hemoglobin E	68.4	1.5 - 3.7%
Hemoglobin E	Present	
Sickling test	Negative	
Protein C	50 IU/dL	Male child 40-60 IU/dL
Protein S	77 IU/dL	Males - Greater than 73 IU/dL Females - Greater than 63 IU/dL
Antithrombin III	90%	80-120%
Prothrombin (PTT)	27 sec	25 - 41 sec
Activated partial thromboplastin time (aPTT):	26 sec	20-40 sec
Echocardiography	normal	
C-reactive protein:	< 1 mg/L	< 5 mg/L
Rheumatoid factor:	< 8 IU/ml	< 25 IU/ml
Anti-double stranded DNA (anti-dsDNA) test	Negative	Positive in Systemic lupus erythematosus Sjögren syndrome mixed connective tissue disease (MCTD).
Antinuclear antibody (ANA)	Negative	Latex agglutination test
Cerebrospinal fluid (CSF) analysis	Glucose: 50 mg/dL. Protein (total): 20 mg/dL. Leukocytes (WBC): 04 /µL . Gross appearance: CSF is clear and colorless.	Glucose: 40–85 mg/dL. Protein (total): 15–45 mg/dL. Leukocytes (WBC): 0–5/µL (adults / children); up to 30/µL (newborns). Gross appearance: Normal CSF is clear and colorless.

IV. Discussion

Moya Moya disease has bimodal age predilection, more common in first decade in Asia and in third and fourth decade in other countries.[1,4] It manifest as transient ischemic stroke in children and aneurysm, transient ischemic attacks, hemorrhage and infarct in adult.[1,4] Typical symptoms at presentations are hemiplegia, paraplegia, headache, convulsions, and involuntary movements.[1,3,4] Hyperventilation induced reduced cerebral blood flow may trigger episodes of transient ischemic attacks causing sudden fall and syncope specially while playing.[3] Thalassemic child has anemia and low hemoglobin level which further causes tissue hypoxia and hypertrophic vascular endothelium leading to micro vascular stenosis.[6-9] Multiple cerebral infarcts and large arterial occlusions in HbE beta thalassemia associated Moya Moya syndrome has also been related to its chronic hypercoagulable state. Hypercoagulable state in thalassemia is multifactorial attributed to endothelial activation, altered platelet function, red blood cell membrane abnormalities leading to activation of the coagulation cascades, and changes in coagulation protein levels.[6-9] Collateral circulation forms from lenticulostriate, thalamoperforating, leptomeningeal, and dural arteries appearing as multiple tortuous flow voids on T1 and T2 weighted sequences. Occlusion of less affected PCA forms pial collaterals, appearing as serpentine and linear hyper intense cortical sulci in T2WI FLAIR images i.e. “ivy sign” due to slow flow in pial collaterals.[1,4] Early radiological diagnosis followed by prompt surgical revascularization improves cerebral perfusion, as well as reduce the risk of subsequent stroke in both pediatric and adult patients.[3]

I. CONCLUSION

Among Asian children Moya Moya syndrome is the most common cause for transient ischemic stroke. Though HbE beta thalassemia is an extremely rare etiological association of Moya Moya syndrome, it is principally emphasized that thalassemia should be considered as one of the etiological association and that children with thalassemia should be screened for future risk of Moya Moya syndrome and transient ischemic stroke.

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