Scholars Journal of Applied Medical Sciences (SJAMS) ISSN 2320-6691 (Online)
Sch. J. App. Med. Sci., 2014; 2(1C):345-347 ISSN 2347-954X (Print)

(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com DOI: 10.36347/sjams.2014.v02i01.0073

Case Report

Vasculopathy in Beta Thalessemia Intermedia : A Rare Case Report

Jayashree Nadkarni[1], Pankaj Pal[2], Rashmi Dwivedi[3], Akhil Singh[4]

[1]Associate Professor, Department of Pediatrics, Gandhi Medical College and Kamla Nehru Hospital, Bhopal, (M.P.) India

[2]Assistant Professor, Department of Pediatrics, Gandhi Medical College and Kamla Nehru Hospital, Bhopal, (M.P.) India

3Professor and Head, Department of Pediatrics and Kamla Nehru Hospital, Bhopal, (M.P.) India

4Senior MD Resident, Department of Pediatrics, Gandhi Medical College, and Kamla Nehru Hospital, Bhopal, (M.P.) India

***Corresponding author**
Akhil Singh
Email: dr_akhilsingh@yahoo.com

Abstract: We report a case of 10 year old thalassemic boy who manifested with sudden onset of left sided hemiparesis. MR angiography revealed bilateral occlusion of internal carotid artery and innumerable dilated and tortuous arterial collaterals giving 'puff of smoke' picture suggestive of Moyamoya disease. Moyamoya disease is an uncommon arteriopathy leading to cerebral ischemia in children and its association with beta thalassemia intermedia is a rarity. This rare but grave consequence of the thromboembolic complications should be considered in these patients. The patient was managed conservatively and has recovered well.

Keywords: Thalessemia, Moyamoya, Stroke

INTRODUCTION

Vasculopathy is any disorder of blood vessel which may result in loss of brain functions due to disturbance in the blood supply to the brain. Moyamoya disease is one of the rare progressive vasculopathyies of an unknown aetiology characterized by a slowly progressive stenosis and obliteration of the large vessels at the base of the brain, affecting mainly the supraclinoid segment of the internal carotid artery and the initial portion of the anterior or middle cerebral arteries and the posterior cerebral arteries1. Moyamoya disease was first described by Suzuki and Takaku, who also graded the disease in 19691. Moyamoya disease commonly involves both intracranial hemisphere but unilateral involvement does not exclude the diagnosis. The name of the disease is derived from the Japanese word moyamoya, which means 'something hazy like a puff of cigarette smoke, drifting in the air' [1]. It is common in Asian population especially in Japan and Korea. Overall incidence is less than one in 100,000 in Japan [2, 3]. It affects females more than males [2]. Peak age of onset is bimodial, with an early peak occurring in the first decade of life and a second peak in the fourth decade of life [3].

Moyamoya disease has been linked to Down's syndrome, neurofibromatosis and sickle cell anemia in past but rarely to β thalassemia intermedia [4-7]. It is an important consequence to be considered especially in patients presenting with stroke. Vasculopathy in brain in Beta – thalassemia cases is rare and may result in thromboembolic complications with poor outcome.

CASE REPORT

A 10 year old boy presented with complaint of weakness of right upper and lower limb, right sided deviation of face and not able to speak properly since 1 day. There was no history of headache, vomiting, fever, trauma, seizures, incontinence of urine or any heart disease. Patient had similar episodes 4 to 5 times in last 7 months but each time recovered spontaneously. He was diagnosed as having β thalassemia intermedia at the age of 3 years and was receiving regular transfusions. He was not admitted for any other complaints in these years.

On examination, patient had stable vitals, all peripheral pulses were palpable and equal, pallor was present, and he had haemolytic facies. There was no icterus, lymphadenopathy, skin rashes or bony tenderness. On systemic examination, patient had right facial nerve palsy, right hemiparesis UMN type, hemiparetic gait and hepatosplenomegaly. Other system examination was not significant.

1.

2. On investigation, haemoglobin was 7.9 gm%. S. ferritin was 2166 ng/ml. The Hb electrophoresis done earlier was suggestive of β thalassemia intermedia (HbA 21.5%, HbA2 3.5%, HbF 72.1%).Electrolytes, LFT, RFT, lipid profile,

Complement 3, ECHO, fundus, Anti Phospholipid Antibodies were within normal limits. X-ray cervical spine showed fused transverse process of C_2-C_3. Contrast CT scan head revealed old infarct in left MCA. In Fig. 1 MRI Brain showed evidence of occluded supraclinoid bilateral ICA with poor visualization of both MCA m1 and m2 segment and poor visualization of a1 segment of both ACA and multiple and innumerable dilated and tortuous arterial collaterals in basilar cistern, both thalamocapsular and capsuloganglionic region arising from the lenticostriate and thalamoperforator arteries, basilar and posterior cerebral artery Significant narrowing of cavernous segment of left ICA was also seen. MRI Brain is giving "puff of smoke appearance" (Fig. 2).

3.

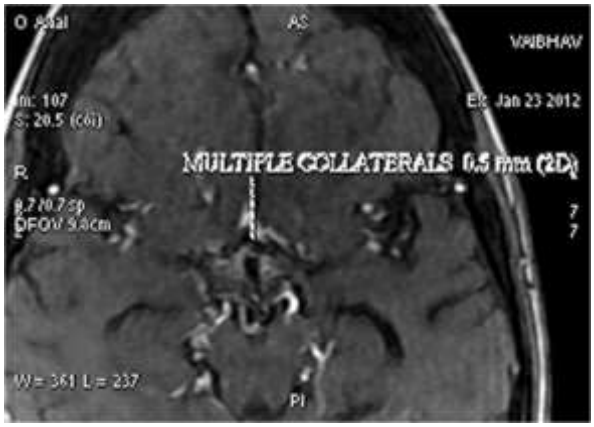
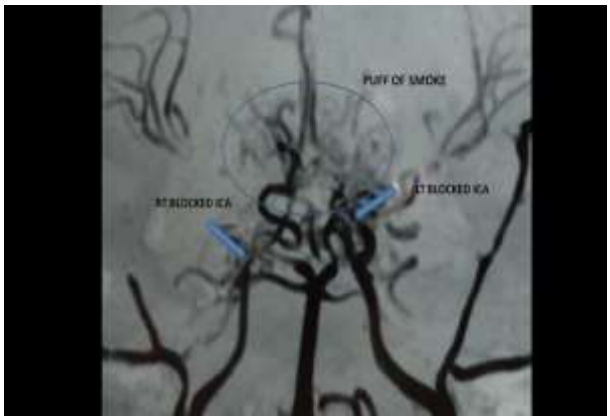


Fig. 1: MRI Brain showed evidence of occluded supraclinoid bilateral ICA with poor visualization of both MCA m1 and m2 segment and poor visualization of a1 segment of both ACA and multiple and innumerable dilated and tortuous arterial collaterals in basilar cistern, both thalamocapsular and capsuloganglionic region arising from the lenticostriate and thalamoperforator arteries, basilar and posterior cerebral artery Significant narrowing of cavernous segment of left ICA was also seen

4.



5. Fig. 2: MRI Brian angiography showing puff of smoke appearance

Patient was managed conservatively and put on T. Pentoxiphyline, T. Ecosprin and T. Defepirone. He showed gradual improvement in power of affected side over a period of 15-20 days and is on regular follow up at our hospital till date. He is doing well and has no neurological deficit till date.

DISCUSSION

Moyamoya disease has a rare association with β thalassemia intermedia which is characterized by steno-occlusive disease of the supraclenoid internal carotid arteries or proximal anterior or middle cerebral arteries, associated with networks of arterial collaterals adjacent to the obstructive lesions. Four cases of this association have been reported in our knowledge till 2010, and the first report of a patient in Turkey [8-11]. Unlike most of the cases of moyamoya disease, our patient responded well with pharmacological management within a period of 15 to 20 days12. Delayed diagnosis and treatment could result in major stroke or intracranial haemorrhage with ensuing permanent loss of functions. Surgical modalities (EDAS/EMS/STA-MCA bypass) should always be considered in case of further progression of symptoms or recurrence of neurological events [13, 14].

CONCLUSION

Association between Moyamoya disease and β thalassemia intermedia though rare is a known entity. Cerebral vasculopathy is a grave consequence of the thromboembolic complications in these patients. With timely diagnosis these patients can be managed conservatively.

REFERENCES

1. Suzuki J, Takaku A; Cerebrovascular "moyamoya" disease: Disease showing abnormal net-like vessels in base of brain. Arch Neurol**.,** 1969; 20(3): 288-299.
2. Kuriyama S, Kusaka Y, Fujimura M, Wakai K, Tamakoshi A, Hashimoto S *et al.*; Prevalence and clinicoepidemiological features of moyamoya disease in Japan: findings from a nationwide epidemiological survey. Stroke, 2008; 39(1):42-47.
3. Wakai K, Tamakoshi A, Ikezaki K, Fukui M, Kawamura T, Aoki R *et al.*; Epidemiological features of moyamoya disease in Japan: findings from a nationwide survey. Clin Neurol Neurosurg., 1997; 99 (Suppl 2): S1-S5.
4. Lamas E, Diez Lobato R, Cabello A, Abad JM; Multiple intracranial arterial occlusions (moyamoya disease) in patients with neurofibromatosis one case report with autopsy. Acta Neurochir., 1978; 45(1-2): 133-145.
5. Koss M, Scott RM, Irons MB, Smith ER, Ullrich NJ; Moyamoya syndrome associated with neurofibromatosis Type 1: perioperative and long-term outcome after

surgical revascularization. J Neurosurg Pediatr., 2013; 11(4): 417-425.
6. Stockman JA, Nigro MA, Mishkin MM, Oski FA; Occlusion of Large Cerebral Vessels in Sickle-Cell Anemia. N Engl J Med., 1972; 287(17): 846-849.
7. Kainth DS, Chaudhry SA, Kainth HS, Suri FK, Qureshi AI; Prevalence and characteristics of concurrent down syndrome in patients with moyamoya disease. Neurosurgery, 2013; 72(2): 210-215.
8. Göksel BK, Ozdogu H, Yildirim T, Oğuzkurt L, Asma S; Beta-thalassemia intermedia associated with moyamoya syndrome. J Clin Neurosci., 2010; 17(7): 919-920.
9. Sanefuji M, Ohga S, Kira R, Yoshiura T, Torisu H, Hara T; Moyamoya syndrome in a splenectomized patient with beta-thalassemia intermedia. J Child Neurol., 2006; 21(1): 75-77.
10. Parker TM, Ward LM, Johnston DL, Ventureya E, Klaassen RJ; A case of Moyamoya syndrome and hemoglobin E/beta-thalassemia. Pediatr Blood Cancer, 2009; 52(3): 422-424.
11. Oberoi S, Bansal D, Singh P, Marwaha RK; Stroke in a young boy with β-thalassemia intermedia secondary to moyamoya syndrome. J Pediatr Hematol Oncol., 2010; 32(7): 568-570.
12. Cantwell JFR, Dettorre RR, Quisling RG, Mericle RA; The use of nitroglycerin for treatment of progressive moyamoya disease: A prospective pilot study of safety and efficacy. J Pediatr Neurol., 2007; 5:101-109.
13. Kim DS, Huh PW, Kim HS, Kim IS, Choi S, Mok JH *et al*.; Surgical treatment of moyamoya disease in adults: combined direct and indirect vs. indirect bypass surgery. Neurol Med Chir., 2012; 52(5): 333-338.
14. Kim DS, Kang SG, Yoo DS, Huh PW, Cho KS, Park CK; Surgical results in pediatric moyamoya disease: angiographic revascularization and the clinical results. Clin Neurol Neurosurg., 2007; 109(2): 125-131.